Juvenile Batten Disease: A Challenge to Conventional Sociological Approaches to Chronic Illness and Disability

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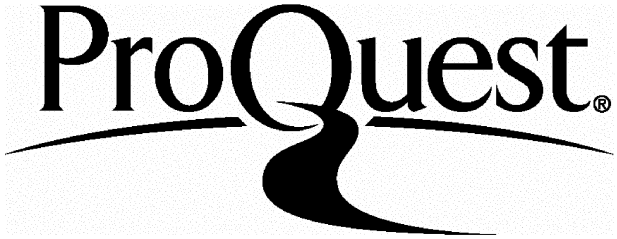

Abstract

Demographic trends towards an ageing population and advances in the treatment of acute conditions and in the efficacy of life preserving treatments have led to an increase in the number of people living with chronic illnesses. These developments, along with the recognition of chronic illness as the most common cause of impairment in the developed world, have raised the profile of chronic illness and the associated issue of disability, making them an increasingly important aspect of the study of healthcare.

This thesis explores the experiences of families of children and young adults living with juvenile Batten disease - a rare neurodegenerative chronic condition - as an exemplar of the changing nature of chronic illness and disability towards the end of the twentieth century. The ongoing debates around definitions and attitudes are examined with particular reference to the symbiotic, and often antagonistic, relationship between biomedicine and sociology in this area.

The experiences of the families of children and young adults with juvenile Batten disease, and the methodological dilemmas inherent in studying a condition of this kind are used to challenge existing approaches to the field. The changing nature of the experience of chronic illness is explored through the growth in community care, self-help and wider access to information. This is related to the sociological literature on chronic illness and disability with

specific reference to the role of users and experts in the research context, the place of the body within the literature, the challenge to biomedicine inherent in both literatures, and the social model of disability. It is suggested that the remit of the sociology of disability needs to be widened to address the embodied nature of disability and to better incorporate disabilities caused by chronic illnesses and rare diseases, profound multiple disabilities and degenerative conditions.

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1 Introduction

Rationale, Concepts, Theory and Research Questions

This thesis sets out to examine the debates around the definitions of chronic illness and disability within the context of young people with juvenile Batten disease and their families and sets out to answer five questions:

1. What is the understanding of chronic illness and disability within sociology?
2. What are the experiences of families living with juvenile Batten disease?
3. Do the experiences of families living with juvenile Batten disease reflect the wider experiences of people living with more common chronic illnesses?
4. Are the experiences of people with chronic illnesses – and in particular juvenile Batten disease – reflected in the current literature and theory around disability?
5. Is it necessary to develop an alternative approach to the theorising of chronic illness and disability to reflect the experiences of the many groups of people with rare chronic conditions who do not appear to be covered in the existing approach?

The chronic illness and disability literatures are explored through the experiences of families with juvenile Batten disease, allowing a unique insight into the adequacy of our existing definitions and understanding in this area.

Juvenile Batten disease is a rare, hereditary, metabolic, neurodegenerative condition, which causes profound motor and cognitive disability and a significantly shortened life expectancy. The symptoms usually develop at approximately the age of six and the average life expectancy of a child with juvenile onset Batten disease is approximately 25. Working within a Critical Realist understanding of the social world, this thesis uses a qualitative ethnographic approach to explore the experiences of families with juvenile Batten disease. These experiences are then used as an exemplar in studying the changing nature of chronic illness and disability and the way such changes are reflected within the relevant literatures. In this introduction I shall outline the details of the four research questions that thesis sets out to answer, and the ways in which the questions are approached in the study. I then set out the rationale for the study and the reasons for using juvenile Batten disease as an exemplar before outlining the key concepts of Juvenile Batten Disease; chronic illness; disability; and Critical realism, and their usage. Finally I shall outline the structure of the thesis itself and how it relates to each of the chapters included.

Research Questions

Five research questions have been addressed within this study with the purpose of looking both at the specifics of day to day life with juvenile batten disease and more widely at the implications that these experiences have for our understanding of chronic illnesses as a whole and, in particular, the crossover between chronic illness and disability, in both experience and literature. Each research question is explicated below along with a brief overview of how the question is being addressed.

1. **What is the understanding of chronic illness and disability within sociology?**

This question is explored through a review of the literature exploring the development and trends within the sociological literature on chronic illness and disability over the last 50 years.

2. **What are the experiences of families living with juvenile Batten disease?**

   - To what extent do the experiences of families with juvenile Batten disease fit the experiences of people with more common chronic illnesses?
   - To what extent is juvenile Batten disease a chronic illness?

- What, if anything, can sociology say about rare conditions?

This set of questions is being addressed predominantly through the data from the family interviews, supplemented by the observational data and documentation collected from schools and care units. The results will then be compared and contrasted with the existing literature on chronic illnesses to identify the extent to which the two experiences coincide. Six key themes of: uncertainty; family relations; stigma; biographical reconstruction and the reconstitution of self; managing medical regimens; and information, awareness and sharing, will be used as the benchmark measurement for experiencing chronic illness. In addition to this the part played by sociology in our understanding of rare chronic illnesses will be assessed in light of previous studies.

3. Do the experiences of families living with juvenile Batten disease reflect the wider experiences of people living with more common chronic illnesses?

- Does the nature of juvenile Batten disease fit in with the challenge to bio-medicine inherent in the sociology of chronic illness?
- To what extent does juvenile Batten disease reflect issues of self help?
- To what extent does juvenile Batten disease reflect issues around styles and locations of care and the people who provide that care?

- Are arguments about the social construction of chronic illness relevant to the experiences of people with juvenile Batten disease?

This set of questions will be answered predominantly through the analysis of the literature and with information collected through the observation of the setting up of the Batten Disease Family Association and the setting up of a care home specifically for people with juvenile Batten disease and other people with conditions that are similar in severity and life expectancy. Particular attention will be paid to the importance of information gathering and the use of mass media and internet resources along with the role played by a range of charities and self help groups both nationally and internationally.

4. Are the experiences of people with chronic illnesses – and in particular juvenile Batten disease - reflected in the current literature and theory around disability?

- To what extent does disability theory reflect issues pertinent to disability caused by chronic illness?
- To what extent does disability theory take into account the particular problems encountered by people with rare chronic conditions and what explanations/solutions are provided for the particular issues these conditions raise?

- What account does current disability theory make of the issues faced by people with profound multiple disabilities and those assisting them with their care?

These questions will be answered through the in-depth exploration of the literature on disability theory in respect to chronic illness and the place of people with chronic illnesses within the literature. Whilst focusing on books and journal articles, information published on the internet and through support groups and disability organisations, is also examined. Government documentation on disability in the United Kingdom and the role given, within it, to the specific needs of people with chronic illnesses is also explored.

5. Is it necessary to develop an alternative approach to the theorising of chronic illness and disability to reflect the experiences of the many groups of people with rare chronic conditions who do not appear to be covered in the existing approach?

This final question pulls together all of the results from the previous section. I am going to suggest that a change, or broadening, in the way chronic illness is approached within sociology is needed along with a reconceptualisation of disability theory to include those who are at present excluded, or at best given lip-service.

Rationale

My purpose in this thesis, then, is to look at the debates around chronic illness and disability through the experiences of families with juvenile Batten disease. The research questions were developed through reading the existing literature and identifying potential problems within it, and, having been involved in research around juvenile Batten disease for the last 5 years, I felt that the experiences of these families exemplified the issues that I wanted to examine.

There are ten issues that are explored throughout the thesis, and through the answers to all four of the research questions, raised through the particular nature of juvenile Batten disease itself, as illustrated in table 1. These issues are built on in each subsequent chapter of the thesis as alternative viewpoints are added.

The experiences of young people and families with Juvenile Batten Disease share both similarities and differences with the experiences of people with more common chronic illnesses. In addition, the previous study that I carried out, on the social care needs of young adults with juvenile Batten disease, allowed me to make links with a number of families and a whole range of professionals working in the area, which enabled me to collect the information that I needed for this study. The timescale, combination of symptoms, severe degenerative nature and inevitable outcome of juvenile Batten's disease make it a very interesting exemplar of some of the most pressing challenges to our existing understanding of chronic illness and disability. As will be examined in more detail later, it is the closures inherent in juvenile Batten's disease that

Table 1

ISSUE RAISED
1. Genetic Disease - Gene Replacement Therapy, The New Genetics, Impact on the Family
2. Rare Disease - Very Limited Literature, Small Sample Population, Methodological Difficulties
3. Diagnosis - Diagnostic Techniques Available,
4. Degenerative Disease – Constantly Changing Reality over a Relatively Short Period of Time
5. Profound Multiple Disabilities - Severe Disability, Affecting Many Aspects of the Individual
6. Cognitive Degeneration – Gradual Loss of Skills, Focus on Maintenance not Learning New Skills
7. Sense of Self - Loss of Means of Maintaining Sense of Self
8. Bio-medicine - Gives Only Hope for Present and Future
9. Specialist Care Needs – Normalisation is not Possible
10. Role of the Family – Carriers, Carers, Proxy Information, Live with the Disease, Continuity

precipitate the challenge to current open theoretical understanding of the area From this point I went back to the literature's on the sociological understanding of chronic illness and disability.

Chronic illness and the associated disability is becoming an increasingly important aspect of the study of health care as the number of people with chronic illnesses increases with the advances in the treatment of acute conditions, the advances in life preserving treatments and the ageing population. Chronic illness is also recognised as the most significant cause of impairment – a survey by the OPCS, for example, identified arthritis as the major cause of disability in Britain (Martin et al, 1988). The latest figures published by the Office for National Statistics, show the proportion of people reporting a long standing illness to have increased from 20% of the population in 1972 to 33% of the population in 1998-99, with a concurrent increase in the number of people reporting a limiting long standing illness from 15% in 1972 to 20% in 1998-99 (ONS 2001). This can, however, be seen as a conservative figure as it relates only to people who class themselves as having an illness that restricts their activities. For example, it is estimated that, in 1984, there were as many as 1 million children in American with severe chronic illnesses and as many as 10 million with less severe chronic illnesses. Taking insulin treated diabetes and HIV as examples, the most recent figures suggest that both illnesses are on the increase. The prevalence of Insulin treated diabetes increase for men and women over the age of 16 between 1994 and 1998 with larger increases amongst the older age ranges (ONS Medicines Control Agency 2001). The trend is similar for incidences of the Human Immunodeficiency

Syndrome with figures from the Public Health Laboratory Service showing diagnosed cases increasing from 2.5 thousand diagnosed cases in 1994 to over 2.9 thousand cases in 1999, and as many as 41 thousand diagnoses of HIV by March 2000 (PHLS 2001).

In addition to the large numbers of people with the more common chronic conditions such as Multiple Sclerosis and Epilepsy, the advances in diagnostic techniques and medical treatment has led to an increasing number of children surviving into adulthood with rare metabolic, genetic conditions. Many of these conditions, such as juvenile Batten's disease, have symptoms which manifest themselves over a prolonged period and result in profound, multiple disability. In addition, the vast majority of these rare, metabolic diseases are not, at the present time, curable, and so, in common with many of the more common chronic illnesses, symptomatic treatment and palliative care are all that can be provided by the medical profession at the current time. It can be suggested that the recent emergence of these conditions into the public arena, through mass media, internet etc, poses a new challenge, initially to medicine and then as a challenge to sociologists studying the nature and impact of chronic illness and disability on society. In addition I am going to argue that studying profoundly disabling, rare conditions in more depth allows us to better understand the links between chronic illness and disability and to identify shortfalls in our current theoretical understanding of chronic illness and disability and the links between the two sets of literature.

Using Juvenile Batten Disease as an Exemplar

There are a number of reasons why juvenile Batten disease is an interesting condition, both in its own right, and as an exemplar of the changing nature of chronic illness and disability. Very little research has been carried out looking at the social implications of living with juvenile batten disease. Two studies have been identified, one carried out in Australia focusing on the impact of juvenile Batten disease on carers (Labbe 1996), and the other being the study of the social care needs of young adults with juvenile Batten disease that I was involved with (Scambler 1999). There is a clear gap in information and informed understanding of the experiences of families living with this type of profoundly disabling, degenerative condition. This study, therefore, at the most basic level, will allow us to build an understanding of some of the key issues relevant to families with juvenile batten disease. In addition to this, an understanding of the social implications of juvenile Batten disease will enable us to better understand other, under-researched, rare conditions of a similar nature.

The limited amount of research available and rarity of juvenile Batten disease itself also results in a concomitantly limited number of researchers working in the area. The role of the researcher is particularly important in this study where I, as the researcher, have also been involved in developing the philosophy of care for a group home for young adults with juvenile Batten disease and am still a member of the committee of the Batten Disease Family Association (see Appendix One for more details). The reflexive approach taken throughout this

research is, therefore, looked at in more detail in the methodology chapter (chapter four).

Moving beyond the exploration of juvenile Batten disease itself, using the disease as an exemplar of rare, chronic, disabling conditions will allow me to examine the chronic illness and disability on a wider level. This allows for an advance in our understanding at both an empirical, experiential level and at a theoretical level relating to the meaning and understanding behind the terms chronic illness and disability themselves. This is particularly important in relation to my research questions which aim to look beyond the experiential level on which much debate around chronic illness and disability is based. There are four key reasons why juvenile Batten disease makes a good exemplar when looking at the changing nature of chronic illness and disability in late modernity. The first is that qualitative research about the effects of rare chronic illnesses has been carried out in very few cases. Juvenile Batten's disease is one of the few conditions where research of this kind, although clearly limited, has been carried out and this gives us a grounding to build on. In addition to this, the fact that I made the initial links with families and professionals in the course of my previous research allows me to access the data more easily.

A further reason why juvenile Batten's disease is a good exemplar is that (as explored in more detail in chapter three) it is an extreme example of the manifestations of a chronic, degenerative, disabling condition over a relatively short time-span. Within the space of twenty years a child will go from being asymptomatic, and often undiagnosed, to being profoundly and multiply

disabled with death as the inevitable outcome. This is combined with the fact that juvenile Batten's disease clearly fits the definition of a chronic disabling illness as it manifests itself over an extended period of time, causes considerable change to a person's lifestyle and experiences of life, and has no cure at the present time. It seems clear from this that juvenile Batten disease is both an informative, and independently interesting exemplar for a study of this kind. The genetics, clinical manifestations and social implications of juvenile Batten disease are looked at in more detail in the next chapter. However, before moving on to look in more detail at the biomedical nature of juvenile Batten disease, it is useful to define the other key terms used frequently throughout this thesis, and to highlight the theoretical, epistemological position underlying this study.

Defining Terms

This thesis will argue (on the basis of the changing nature of chronic illness as a category, and that chronic illness and disability, although clearly closely related in practice are, on the whole, treated as separate entities within theory) that the concepts of chronic illness and disability are in need of re-classification. However, in making this argument it is important to state the foundations from which any new understanding of the concepts of chronic illness and disability will emerge. Thus it is necessary to state the definitions of chronic illness and disability that are going to be used, explored and challenged throughout this thesis, to highlight the key aspects of juvenile Batten disease itself, and to

outline an understanding, through theory, of the type of society/world/reality in which all of the concepts and experiences are being used.

Juvenile Batten Disease

Juvenile Batten disease is the juvenile onset form of a group of diseases known as Batten disease, all of which have similar symptoms, although with different time scales and causes. Juvenile Batten disease is a genetic, metabolic disease which develops at approximately the age of 6 and results in a life expectancy of approximately 20 years from symptom onset. The key symptoms of juvenile Batten disease include epilepsy; severe visual impairment; cognitive degeneration (including the loss of short term memory and the loss of daily living skills); motor degeneration (starting with the loss of fine motor skills and ending with the extreme limitation of controlled movement); loss of speech and verbal communication skills and psychological and behavioural problems. All of these losses occur over a twenty year period resulting in a premature death. There is currently no cure for juvenile Batten disease although genetic research is looking into the possibilities of genetic treatments, and a mouse model of the disease has been developed by a team of researchers at University College London. The genetic inheritance of juvenile Batten disease, along with the clinical manifestations, diagnosis, symptomatology and treatment are all looked at in more detail in chapter three, but this gives an idea of the impact that the disease has on the lives of the children who have it, and on the lives of their wider families.

Chronic Illness

At the current time there is little debate over the nature of chronic illness as a category. In the context of this work the term 'chronic illness' is taken to refer to various conditions which have a long-term impact on the lives of the sufferers. Chronic illness covers a huge range of conditions:

> It includes congenital and acquired physical impairments, mental illness and mental impairment (although its medical status is questionable), stable and progressive (including terminal) conditions, temporary and permanent injuries, and a variety of 'disfiguring', although not always functional, impairments (from birthmarks, burns and scars to obesity and thinness). (Barnes et al, 1999, pp48-9)

Chronic illnesses are generally illnesses where treatment for the underlying condition is not available. The symptoms of chronic illnesses are, however, often treatable, although the emphasis is on enhancing and sustaining quality of life rather than irradicating symptoms.

Disability

The term disability has been defined in a variety of ways over the past quarter of a century alone, and, although the definitions in use will be analysed in more detail in chapter seven, it is useful to outline the two main positions from the outset, and to highlight the latest attempt to reach a middle ground between the two extremes. The 'International Classification of Impairment, Disability and

Handicap' (ICIDH), adopted by the World Health Organisation in 1981, is the most commonly used definition of disability amongst medical professionals and in a wide range of fields outside of disability theory itself. ICIDH states that:

> **Impairment** - an impairment is any loss or abnormality of psychological, physiological or anatomical structure or function.
>
> **Disability** - a disability is a restriction or lack (resulting from an impairment) of ability to perform an activity in a manner or within the range considered normal for a human being.
>
> **Handicap** - a handicap is a disadvantage for a given individual, resulting from an impairment or a disability, that limits or prevents the fulfilment of a role that is normal (depending on age, sex and social and cultural factors) for that individual. (WHO 1981)

Although this is still the most commonly used definition of disability, the ICIDH has been widely criticised for placing the emphasis on the individual and their impairment, and not on society or the environment. The social model of disability has developed alongside the ICIDH as an alternative model, which suggests that disability is caused solely through the inadequate adaptation of the environment to the needs of people with impairments. The clearest version of this definition of disability, which is in wide use in the areas of disability studies and politics is as follows:

Impairment - lacking part or all of a limb, or having a defective limb, organ or mechanism of the body.

Disability - the disadvantage or restriction of activity caused by a contemporary social organisation which takes no or little account of people who have an impairment (whether it is physical, sensory or intellectual) and thus excludes them from participation in the mainstream of social activities. (adapted from UPIAS, 1976)

Again there are criticisms levied at the social model of disability, most centring on the removal of biological impairment from the equation and the subsequent gifting of the body to the medical establishment unchallenged.

From 1996, the World Health Organisation has been working – in consultation – to develop an up to date version of the ICIDH, combining aspects of the medical and social models of disability, and providing a tool for uniform measurement of disability across the world. In the last year the 'International Classification of Functioning, Disability and Health' (known as ICIDH2) has been released. ICIDH-2 is a proposed scale composed of measures of body functions and structures, Activity, Participation and Contextual factors, which has been designed to incorporate aspects of the social model of disability, and to include environmental factors. It has also been designed with the aim of utilising neutral terminology and avoiding past stereotypes and negative connotations. At present ICIDH2 is best described by the WHO Information office as follows:

The overall aim of the ICIDH-s classification is to provide a unified and standard language and framework for the description of human functioning and disability as an important component of health...ICIDH-2 organizes information according to three dimensions: body level; individual level; and society level.

1. The Body dimension comprises two classifications, one for functions of body systems, and one for the body structure.
2. The Activities dimension covers the complete range of activities performed by an individual.
3. The Participation dimension classifies areas of life in which an individual is involved, has access to, and/or for which there are societal opportunities or barriers.

A list of environmental factors forms part of the classification. Environmental factors have an impact on all three dimensions and are organised from the individual's most immediate environment to the general environment. (WHO Press Release no. 19, August 1999)

In the context of this study the term disability is going to be explored in detail and the alternative definitions are going to be put to the test using the exemplar of juvenile Batten disease and the multiple, profound disabilities that it precipitates. Until such a time as an alternative definition is developed,

disability is taken to refer to the profile of activities available to a person with one or more impairments.

Critical Realism

In the words of Sayer (2000: 2) "the defining feature of realism is the belief that there is a world existing independently of our knowledge of it". This reflects Bhaskar's assertion that the world cannot simply be reduced to what we 'know' about the world, the conflation of what 'is' with what is 'known about' is what Bhaskar terms the 'Epistemic Fallacy' (Bhaskar 1989). He suggests that a common mistake within social research is conflation of the ontological with the epistemological. In justification for this position, Sayer argues that it is the evident fallibility of our knowledge that discounts the possibility that the world exists through our knowledge of it, for, this being the case, our knowledge of the world could be nothing but infallible. Thus, the very fallibility of our knowledge of it predisposes the existence of the world.

From this starting point the critical realism developed by Roy Bhaskar (1978) suggests that the 'social and natural world' consists of three domains, the 'empirical', the 'actual' and the 'real'.

Empirical the domain of experience, founded on observable events, although as Bhaskar points out, not all events are in fact experienced.

Actual this domain contains both events and experiences.

Real this domain contains generative, causal mechanisms which are independent of our knowledge of them. These are the deep underlying events or causes within the social and natural world which are often not experienced but are nevertheless real events, experiences or mechanisms.

Using these domains, it can be suggested that, for example, the body, the self, or society have an ontological depth which is independent of epistemological claims, right or wrong, as to their existence. Thus the embodied nature of human beings constrains what they are, who they can become and what they can do (Scambler & Higgs 2000).

This is a brief outline of the essence of a 'critical realist' ontology from which we are seeking to understand the changing nature of chronic illness and disability through the specific example of juvenile Batten disease. Taking a broadly 'critical realist' theoretical stance allows us to look at the direct, observable, experiences and events effecting families living with juvenile Batten disease whilst, at the same time, taking these experiences beyond the surface level to look at the ways in which they fit in with, and challenge our whole theoretical approach to the study of chronic illness and disability within sociology. This study does not just focus on experience, it goes from the empirical to the actual to the real and back again. We are suggesting that current debates have become sterile and that juvenile Batten disease is about more than just experience. For a more detailed explanation for the reasoning behind the adoption of this particular theoretical stance, please see chapter four.

This is a brief outline of the key concepts which are being used in this thesis. More details on the debates surrounding the definitions of disability and critical realism that I have highlighted can be found in the literature review in chapter two, an exploration of the social model in particular, and it's relevance to the experiences of families with juvenile Batten disease and other similar conditions can be found in chapter seven, and further details of the critical realist approach and the reasons for using it in this study can be found in chapter four.

Thesis Structure

Having set out the research questions, rationale, and definitions of the key terms being used in the study, the last thing to note is the structure of the thesis itself and the way in which the research questions have been incorporated into an argument. The thesis contains eight chapters, reflecting the research questions around which the study has developed. This chapter lays out the initial reasoning behind the project, and examines the questions which are being addressed. The second chapter incorporates the literature review, establishing the development of the sociology of chronic illness and disability and examining the variety of theoretical approaches that have been adopted within this area, and introducing the debates that will be looked at in more detail in later chapters. The third chapter then looks in more detail at juvenile Batten disease itself; its genetic heredity, symptoms, treatment, and current research developments. Chapter four focuses on the specific methodological issues

surrounding the study of juvenile Batten disease, problems that have been encountered, and the data collection techniques used within this study.

The four remaining chapters address each of the research questions in turn: so chapter five addresses the experiences of families living on a day to day basis with juvenile Batten disease; chapter six looks the changing nature of such experiences over recent years, particularly in relation to the development of information technologies and changes in ideologies of care; and chapter seven focuses on chronic illness within the disability literature. Chapter eight then concludes with a discussion of the wider issues surrounding sociological theory pertaining to chronic illness and disability and the relevance of current theory with respect to the increasing number of rare, disabling chronic illnesses with life expectancies reaching adulthood and beyond.

2 A Review of the Literature on the Sociology of Chronic Illness and Disability

As established in the previous chapter, this thesis sets out to examine the debates around the definitions of chronic illness and disability. This necessitates a detailed look at the two literatures and the themes, theories and approaches that have developed through them. This chapter, therefore, looks in some detail at sociological approaches to the study of chronic illness and disability and at the changing conceptualisation of these concepts through the development of the area. This review looks specifically at the development of the literatures and does not seek to relate the findings to the literature on juvenile Batten disease, as this is done both in chapter three and chapter six where we look in more detail at the details of juvenile Batten disease itself, and specifically at how it fits in with the literature.

This chapter starts with an overview of the major theoretical approaches to the sociological study of chronic illness and disability as it has developed before looking at the studies that have been carried out and some of the key themes that have emerged from them. It then looks in some detail at the historical development of the concept of disability and at the key debates surrounding the

definition of the term before finally looking at the most recent developments reassessing the relationship between disability and the body, and the ongoing debate about disability and the new genetics.

The Sociology of Chronic Illness

The sociology of chronic illness emerged in the 1950's when the American sociologist Talcott Parsons (1951) first focused sociological attention on to health and illness. Although there is little disagreement as to the definition of chronic illness, as stated in the introduction (chapter one), a variety of different approaches have developed over the past half a century suggesting different ways of looking at and understanding the experiences of people with chronic illness, focusing specifically, in later decades, on the day to day experiences of living with a condition of this type. This review focuses on the multitude of theoretical approaches that have been utilised since the seminal work of Parsons. The key themes that evolved through the body of research focusing specifically on the experience of living with a chronic illness on a day to day basis are then highlighted in more detail.

Theoretical Approaches

A variety of theoretical approaches have been adopted throughout the history of the sociology of chronic illness, from the initial focus on the sick-role in the 1950's through to the recent summaries of the interpretativist research on the day to day experiences of people with a whole range of chronic illnesses that

have been compiled in the late 1990's. This section looks in some detail at the key approaches that have been taken and the resulting themes and wider understanding.

The Functionalist Approach

The sociology of chronic illness developed through the realisation that the bio-medical model of health does not adequately account for the significant effect that chronic illness has on the lives of the sufferers and their families. The area was introduced to sociology through the publication of 'The Social System' (1951) in which Talcott Parsons looked at health and illness and the maintenance of social roles. Parsons work comes from a functionalist perspective suggesting that the maintenance of social roles is necessary to keep society running smoothly. He suggested that health is the norm and illness is abnormal, and therefore that a person who is unwell is deemed to be deviant. People who are unable to work through illness are seen to need legitimisation for their inability to perform their 'normal' social roles and so they take on a 'sick-role' for a temporary period until they are able to revert to their usual roles. Parsons sees the 'sick-role' as a form of 'sanctioned deviance' which encompasses both rights and duties. The person who is ill has to seek medical help, comply with any medical suggestions and actively want to return to full health. In return they are given temporary legitimisation for not continuing their normal roles and are not held responsible for their illness or expected to become healthy again unaided by biomedical science.

Parson's suggested, further, that medical power is needed to legitimise the temporary withdrawal from mainstream society and that the situation works to the advantage of both the doctor and the patient as they have the common goal of wanting to return health to the patient. Thus, medicine can be seen to have an important role in social control through legitimising and monitoring the course of illness and legitimised deviance in society. This theory of the social implications of health and illness introduced the study of the sociology of health and illness.

Parson's work has been widely criticised within the field of chronic illness. It has been suggested that his model fails to take into account people with long term or chronic conditions who are forced to remain in the 'sick-role' for extended periods of time, if not permanently, and for whom the 'sick-role' becomes their normal state, forcing them to accept the duties that accompany it (Kassebaum & Bauman, 1965). In addition to this, work by Friedson (1970) looks at Parson's view of the acceptance of a 'sick role' by society. Friedson suggested that the level of acceptance of the legitimisation of the 'sick role' depends on whether the illness is seen as serious, whether the individual is seen as responsible for causing the illness and on whether the illness is accepted as a legitimate one.

The idea of a sick role was added to by Gordon who advanced the idea of an 'impaired role' that is permanent and extremely difficult to move out of (1966). Building on this Sieglar and Osmond (1974) in their work on madness suggest that having an 'impaired role' forces people into an identity as a 'second class

citizen'. In the book 'Exploring Disability' Barnes et al suggest that this view is perpetuated through the focus on adaptation and normalisation and the biomedical approach to the treatment of impairment and chronic illness. They suggest that people with chronic illnesses are thus 'treated more as objects than as active participants in the treatment process' (Barnes et al, 1999, p42).

The Interactionist Approach

The idea of illness as deviance was taken and developed by sociologists working from a symbolic interactionist perspective who developed the labelling theory looking at the processes through which a person is labelled as ill and deviant. The labelling theory was most comprehensibly applied to the area of health and illness through the study of stigma carried out by Erving Goffman (1968). Goffman studied the categorisation of people as 'normal' or 'abnormal' in society through interaction. He identified three classes of stigma covering: 'abominations of the body - the various physical deformities' - including things like homosexuality, paraplegia and dwarfism; 'blemishes of individual character'; and 'the tribal stigma of race, nation and religion'. The negative image that these groups carry is seen to be contagious and is transferable to family and friends who are said to acquire 'courtesy stigma' (p 44). When looking at stigma Goffman focuses on the 'know-about-ness' of the condition. He distinguishes between discredited and discreditable status. Discredited conditions - such as acute psoriasis - are visible and immediately apparent on interaction and the focus of the interaction is on managing the tension of the reactions of 'normals'. Discreditable conditions - such as

controlled epilepsy - are where the condition is not immediately apparent and the issue for the person is the management of information and decisions on who to inform about the diagnosis.

Goffman goes on to distinguish three ways in which people cope with the stigma of a chronic illness. These are 'passing', 'covering' and 'withdrawal'. 'Passing' is where the focus is on management of information and the person is discreditable, 'covering' is when a person is discredited and focuses on the management of tension, and 'withdrawal' is where the person removes themselves from any interaction with 'normal' people in order that they do not have to cope with the problems of interaction. He suggests, further, that the stigmatised person is then forced into a role as an 'abnormal' person and all other aspects of their life have to fall within this role. The use of stigma roles can be seen as a way of maintaining control of people who do not fit neatly into broader society. It is expected that the stigmatised people should make a concerted effort to 'fit' back into society in their new roles and if they are seen to adjust well then they are acknowledged. 'This means that the "good adjustment" of the handicapped is actually a quality granted to them by others. Then people say about them things like, "he's very brave", or "she's always so cheerful"' (Radley, 1994: p154).

The work of Goffman is criticised by writers who challenge his view that chronically ill people have no choice but to accept their negatively stigmatised roles within society. As Barnes explains:

> "For critics, Goffman's account is obsessed with the defensive, anxiety-ridden and largely doomed manoeuvrings of stigmatised individuals, and of their acceptance of the negative label" (Barnes, 1999, p47).

Higgins (1981) studied the management of stigma by a group of deaf people. He found that the emphasis was placed on maintaining everyday functioning rather than managing their stigmatised identity. A number of studies looking at the effects of stigma on people with learning disabilities also found that the negative images were actively discarded (Bogdan & Taylor, 1989; Booth & Booth, 1994). Goffman's focus on negative stigmatisation was further challenged through research on Epilepsy (Scambler and Hopkins, 1986; Scambler, 1989) which found that actual occurrences of negative stigmatisation - 'enacted' stigma - were relatively rare, and that the fear of 'enacted' stigma - 'felt' stigma - was significantly more common.

The Conflict Approach

The input of conflict theorists into the sociology of chronic illness and disability focuses in particular on power relations between the medical profession and the lay population. Waitzkin (1989) suggests that the relationship between the doctor and the patient is a simple a case of a medical interaction but can also be seen to have an important role within the wider social context. He states that often when people with a chronic illness or disability consult a doctor it is for non-medical reasons, such as a doctor

examining an employee for benefits or sick leave authorisation. Silverman (1987) follows on from this to suggest that chronically ill patients with an expertise acquired through experience are one of the groups that are beginning to challenge the power of the medical profession.

The Interpretative Approach

Interpretative studies are possibly the most widespread development in theoretical approaches to the study of chronic illness and have provided a fresh input into the area. Interpretative studies focus on the ways in which people understand health and illness and the concomitant interactions between the individual and society (Radley, 1994). The social disadvantage accredited to people with a chronic illness and/or disability is seen as resultant of the ways in which the individual interacts with society and vice versa (Williams and Wood, 1988). The Interpretative approach focuses on the complexities of the subjective experience of chronic illness in everyday life.

The interpretative, experiential approach developed from the understanding that people with chronic illnesses are interdependent members of a social network. The focus of research, thus, must be on the experiences of the family as a whole rather than just those of the individual and on the way in which the individual interacts with society. Experiential research looks at interaction and negotiation (Strauss and Glaser, 1965; Roth, 1963), coping strategies, the cultural and organisational demands placed on the family, and the social factors which may influence the experience such as gender, ethnicity, age and stage in the life

cycle. All aspects of life with a chronic illness are assessed from the perspective of the individual and the family, looking at the subjective responses to constraints at home, at work and in all other aspects of life experience. The final focus of this approach is to look at the long-term impact of chronic illness on people's lives and the way in which priorities, problems and strategies change as the disease process progresses.

Advocates of this approach suggest that there have been many studies looking at structural issues such as medicalisation, the doctor/patient relationship, use of services and so forth but that these studies have failed to look at specific illnesses. In addition to this it is suggested that broad studies on disablement often fail to look at the subjective experiences of the people with the disabling conditions (Anderson & Bury, 1998). These criticisms have been answered through the development of a body of research focusing on the experiences of people living with chronic illnesses. There have been a large number of studies, over the last 30 years, looking at the subjective experience of living with specific chronic illnesses including: Epilepsy (Ziegler, 1981; Schneider & Conrad, 1983; Scambler & Hopkins, 1986; Scambler, 1989, 1998), Multiple Sclerosis (Acheson, 1985; Baum & Rothschild, 1981; Strauss, 1984; Robinson, 1988, 1998), Parkinson's Disease (Singer, 1973, 1974; Pinder, 1998), Rectal Cancer (Macdonald, 1998), Psoriasis (Jobling, 1998), Arthritis (Bury, 1998), Stroke (Anderson, 1998), Tuberculosis (Roth, 1963) and, more recently, HIV and AIDS (Aggleton et al 1989, Green and Platt 1997). The findings of these studies and others are looked at in some detail when we explore the themes that have emerged through this research.

The Social Constructionist Approach

The social constructionist approach to the sociology of chronic illness is concerned with the body. Stemming from the work of Foucault (1965, 1976) which looks at the changing nature of disease through the different ways in which the body has been understood throughout history, the gaze has focused on the increased importance of the 'body' in a consumer culture, the emphasis on looking good and the implications of this for the disabled body:

> The rise of commodity culture to prominence in western societies has resulted in the ageing body and the disabled body becoming sources of great anxiety. A body that does not function 'normally' or appear 'normal', that is confined to a wheelchair or bed, is both visually and conceptually out-of-place, as evidenced by the lack of public facilities for people with disabilities or the elderly. (Lupton, 1994: p38)

At the same time the body, now required to be perfect, is provided with all kinds of opportunities for improvement from medical technology to an endless stream of diet and fitness regimes.

There is a clear understanding throughout this literature that weak, unattractive or disabled bodies are rejected or avoided as distasteful. Healthy, 'able' bodies are seen as morally superior (Elias, 1978). Nettleton (1995) highlights the

implications of this in overcoming shame and embarrassment both for disabled people and those who assist them with intimate or personal care. In addition the work of Sacks (1984), Charmaz, (1987) and Mathieson and Stam (1995) identify the effect that the onset of illness has on body consciousness and the awareness that the body is not functioning in the way that is expected of it. Barnes et al (1999) see the social constructionist approach as disregarding the biomedical view of the body and refocusing on the way in which the body is created through discourse and language. Thus the impairment becomes secondary to the effect it has on discourse.

Key Themes Within Our Current Understanding of Chronic Illness

A whole range of features are common to the experience of chronic illness in everyday life. As identified by Turk (1979) these features provide a common framework of understanding around what it is to have a chronic illness.

> "Despite the range of conditions and disabilities encompassed by the category, a number of common features are concomitants of each illness. All chronic illnesses represent assaults on multiple areas of functioning, not just the body. Patients with various chronic illnesses may face separation from family, friends, and other sources of gratification; loss of key roles; disruption of plans for the future; assault on self-images and self-esteem; uncertain and unpredictable futures; distressing emotions such as anxiety,

> depression, resentment and helplessness; as well as such illness related factors as permanent changes in physical appearance or in bodily functioning." (in Sarason & Spielberger, 1979, p291).

These common features appear time and again in the many studies that have been carried out.

The six key themes which are common to many of the studies on chronic illnesses and disability are: uncertainty, family relations, stigma, biographical work and the reconstitution of self, managing medical regimens and information, awareness and sharing. An exploration of these themes in some detail ,illustrated through the many studies that have been conducted, clearly delineates the shape of our current understanding and definitional framework around what it is to have a chronic illness.

Uncertainty

The first common theme is that of uncertainty, As identified by Turk, this relates not just, in many cases, to an uncertain aetiology, but also to an uncertainty about the trajectory of the disease and the future of the person with it. In her work on Parkinson's disease, Pinder (1990, 1998) identifies the importance of uncertainty in the daily management of the lives of people with this disease. There is both an uncertainty of aetiology and of disease trajectory. In addition to this some of the symptoms experienced stem from the disease, some may be related to the high levels of drugs taken to control the symptoms of the disease and others may be both disease and drug related symptoms. The

dual uncertainty of both aetiology and trajectory can be similarly found in the experiences of people with multiple sclerosis (Robinson, 1998) and arthritis (Anderson, 1998) where it is difficult to know from day to day how symptoms will effect people and what activities they will and will not be able to undertake. This uncertainty and the profound effect it has on daily life was recognised as early as 35 years ago before the breakthrough in information on Tuberculosis (Roth, 1963). Research is also beginning to emerge looking at the disease trajectory of HIV and AIDS patients suggesting that uncertainty plays a large part in the management of their long-term life objectives. People with HIV do not know how long they will remain a-symptomatic before they develop AIDS or if they will develop AIDS at all, and if they do develop AIDS there is uncertainty about how long they will have the syndrome before they die (Aggleton and Thomas 1988).

It has been shown that, in the case of some of the chronic illnesses where uncertainty is predominant, the diagnosis can be welcomed as a legitimisation of experiences (Robinson, 1988). The legitimisation of the sick-role can also alleviate the guilt over not being able to perform everyday tasks (Badura and Waltz, 1984; Mayou, 1986; Brown et al, 1981). In addition, it is suggested that, even where the end result of the condition - i.e. slow degeneration and death - are known, there may still be a great deal of uncertainty and unpredictability as to when and how degeneration will occur (Hobbs and Perrin, 1985).

Family Relations

The second theme is that of family relations. Chronic illness can be seen to affect the whole family and not just the person who has the illness. The reactions and support of a family can influence a person's response to their illness and subsequent outcomes (Kasl, 1983). The most common way in which families are affected by chronic illness is through the need to care for the person with the illness. Families are seen as having a key role both in the care and the rehabilitation of the chronically ill person (Smith, 1979; Anderson, 1987, 1998). The majority of carers of people with chronic illnesses are unpaid (usually female) family members who often need to pay constant attention to their loved one and may suffer from exhaustion, social isolation, depression and financial hardship (Fitzpatrick, 1998). Anderson (1998) in a study on stroke victims reiterated the fact that the family are the key carers, suggesting that caring for a chronically ill family member causes additional distress through the loss of the person as an active social partner and through the demands placed on a carer which reduce time available for social activities.

Anderson and Bury suggest, further, that a chronic illness can have a more significant effect on family life and roles if the woman is ill because this can cause changes in traditional domestic roles and have a huge impact on the life of the family unit (1998). In addition to this, chronically ill women are more likely to be abandoned and more likely to live longer and be alone when they experience chronic illness (Anderson & Bury, 1998). In a previous study Grove (1973) found that people who live alone with a serious illness suffer because they lack social support, informal support and domestic care.

Anderson and Bury also identify a potential discrepancy between the ways in which the person with the condition and their family view the seriousness of the illness, suggesting that the families may see the condition as far more disabling than the person who has it does or, vice versa.

In their study on the experience of living with epilepsy, Scambler and Hopkins (1998) found that both the stigma of having the condition and the occurrence of seizures affected the equanimity of the family. It was also stressed that the needs of families fell beyond the scope of bio-medical services and that wider provision needed to be made to assist the families of people with chronic illnesses (Kanthor et al, 1974; Palfrey et al, 1980). This is reflected in the work of Burr (1985) who suggested, for example, that the impact of the chronic illness moved far beyond biomedical needs to the extent that career aspirations of parents of children with chronic illnesses can be adversely affected by the need to provide care and access specialist health care. It is widely recognised that chronic illness has an impact on both finances and employment, effecting both immediate costs and long-term employment prospects (Conover, 1973; Townsend, 1979; Whitehead, 1987; Royal College of Physicians, 1986). In addition to the above, time is also a commodity affected by chronic illness and families often felt deprived of leisure time, time for activities with the family and time to spend with their partners (Burr, 1985).

Stigma

As mentioned in the previous section, the third theme stemming from the research on chronic illness is that of stigma. Anderson and Bury suggest that

the potential spoiling of identity through the negative reactions of others to changes in normal bodily or behavioural experiences is at the centre of much of the misery of chronic illness (1998). Scambler (1984) in his study of people with epilepsy, found that the very act of being labelled as epileptic caused great distress. It was clear from his study that the fear of the acquisition of a label challenged people's image of themselves as 'normal'. He also found that the fear of stigma, 'felt' stigma, was far more predominant than actual acts of stigmatisation, 'enacted' stigma. This concept was also recognised by Macdonald who referred to it as 'perceived' stigma (1998). Moving on from this, Schneider and Conrad (1981) identified adjusted and unadjusted modes of adaptation to the new identity, ranging from fully adjusted people to the pragmatic, secret and quasi-liberated reactions. In a series of interviews of people who had undergone an ileostomy, Kelly (1992a, and 1992b) identified the difficulties on managing the private and public identity, coping with the private changes in bodily function and image along with public perceptions and stigmatising attitudes.

Cornwell (1984), looking at working class perceptions of health found a link between perceptions of health and of 'moral worth', a concept which is clearly visible in later work on HIV and AIDS (Aggleton and Thomas, 1988; Aggleton et al, 1989, Green and Platt 1997) which recognises the popular view of some chronic illnesses as linked to a low level of morality. In her work on 'dread diseases', Susan Sontag (1991) highlights the public perceptions of diseases in society and the moral judgements associated with different conditions, such as fear, pity and contempt. A number of studies suggest that the negative

perceptions held about certain illnesses exacerbate the, already problematic and unpleasant, experiences of people with chronic illnesses (Scambler and Hopkins, 1986; West, 1979; Schneider and Conrad, 1983). Albrecht et al (1982) also found that people distinguished between symptoms affecting the mind and those affecting the body suggesting that more stigma was faced by those with mental symptoms than physical ones. In his study of Psoriasis, Jobling (1998) points out the need for more research in to the stigma faced by people with highly visible skin diseases where the unfounded fear of contagion is also an issue

Biographical Work and the Reconstitution of Self

Research shows that the diagnosis of chronic illness has a huge impact on the self-image of the diagnosed person, as well as on their public image, and causes them to rethink their past, present and future. Bury (1997) suggests that there are two ways in which this type of biographical disruption can be overcome, firstly, through the construction of an account of what has happened and why and, secondly, through seeking legitimisation for the condition and the lifestyle changes it necessitates. In contrast, Bloom, through his analysis of the experience of a woman with kidney disease and cancer (1992) charts a progression from the 'dys-embodied self' through reflexive self-awareness to a re-alignment. On a cautionary note Williams (1996) points out the precarious, fragile nature of such negotiated settlements and the need for constant readjustment and negotiation. Following from this Pinder (1998) suggests that meanings change over time as symptoms change and thus new negotiated settlements need to be reached.

The importance of the negotiation and renegotiation of identity through discourse has been identified through a number of studies. Locker (1983) identifies the need for the patient to simultaneously cope with the biographical and social implications of having a long-term condition. In addition Anderson and Bury (1998) suggest that people with a chronic illness redefine their health status to incorporate their disability, and thus that it is possible to be both healthy and to have a disabling condition. This mirrors earlier work by Herzlich (1973) and Blaxter (1990) which suggests that lay definitions of health may often be more to do with feelings of wellbeing and functional ability than with the presence or absence of disease. However, the wide spread feelings of social isolation experienced by many people living with chronic illness, and by those who care for them, are testimony to the difficulties experienced by many people in adjusting to a new and different identity, image and lifestyle (Dartington et al, 1981).

Managing Medical Regimes

The importance of managing the medical regime is looked at in detail in the work of Pinder (1998) on living with Parkinson's disease. She highlights the need to balance the control of symptoms with the management of complex drug regimes and the need to cope with both the symptoms caused by the disease and the additional symptoms caused by the drugs taken to control the disease. In his work on Psoriasis, Jobling (1998) goes on to describe the way in which the treatment regime can come to dominate the everyday lives of people living with the condition and the need to balance the benefits and costs of the treatment.

He also highlights the need for negotiation between the doctors and patients as to the regime followed and how it should be managed. Following on from this, Locker (1983) identifies the fact that time spent on managing a complex medical regime cannot be spent on other, more enjoyable things.

Information, Awareness and Sharing

Research over the last two decades challenges the view of the doctor/patient relationship, as characterised through the work of Parson's on the 'Sick-Role', in terms of the dominant and subservient participants. Fitzpatrick (1990) suggests that, for chronically ill people, there is a higher level of dissatisfaction with doctor/patient relationships than with any other aspect of care. Patrick et al (1983) go on to suggest that there is widespread dissatisfaction with the explanations of causes of disease or health problems given by doctors. Harding and Modell (1985) found that nearly 50% of asthmatics interviewed were dissatisfied with the communication that they received from their doctors. These findings were also reflected in work carried out by Cooper and Huitson (1986), who found that a large number of patients with epilepsy, in their audit of 30 general practices, felt unable to discuss general worries with their doctor. Ziegler (1981), also in a study on epilepsy, found that doctors were preoccupied with the management of the condition and disinterested in other aspects of care. He suggests that doctors must see patients as potential 'clinical partners' and be willing to share information of the best care is to be provided. This viewpoint is echoed by Schneider and Conrad (1983) who suggest that doctors and adult patients need to be co-participants in care.

The importance of the quality of information and support that are provided was highlighted by Macdonald (1998) in her study of living with rectal cancer, in which she suggests that the quality of information provided both pre and post-operation may affect people living with rectal cancer throughout the rest of their lives. In addition she suggests that an improvement in the quality of information and communication would play a significant part in the reduction of the stigma currently attached to rectal cancer. The lack of information available on practical or emotional issues is highlighted by Anderson (1998) who states that the professionals who play the key role in disseminating information are bound up in the physical impairment and issues around rehabilitation and do not consider the need for other types of information. This is reflected in the fact that there is a shortage of information on the practicalities of day to day care of someone with a chronic illness (Burr, 1985).

In addition to the themes illustrated above, the importance of issues such as quality of life (Fitzpatrick, 1990, Anderson, 1998), caring and Normalisation (Strauss,1984) have also been established. The double disadvantage of being a woman with a chronic illness has also been highlighted. Changes in domestic roles are more likely to occur if a woman becomes chronically ill, women are more likely to be abandoned and are also more likely to live longer and be alone when they become chronically ill (Whitehead, 1987). In addition, the focus, in contemporary society, on the performative element of society, the active, skilled professional and consumer with an emphasis on interpersonal skills, performance and self-control makes it increasingly difficult for people

living with chronic illness to successfully 'normalise' themselves as is expected (Anderson and Bury, 1998).

Many of the themes that I have highlighted in relation to the literature on chronic illness can also be found within the disability literature. The concept of 'normalisation', for example, has played a key role in the lead up to current debates around disability politics and the identity of disabled people. These debates are looked at in more detail in the second half of the chapter where I shall look the development of disability studies and the body of work that has been carried out in this area.

The Sociology of Disability

Current estimates suggest that approximately 10% of the world's population, more than half a billion people, are disabled. It is predicted that this number will rise dramatically in the next quarter of a century (International Disability Foundation 1998). The area of disability studies has developed alongside an increasing awareness that 'disability' is about more than the care of functionally impaired people. In reviewing the sociological literature on disability we will start with a historical overview of the changing understanding and treatment of disability up to 1960's when a distinct body of literature developed examining the issue of 'disablement' and the experiences of people living with disability. Central to this literature is a large body of work around the defining of the concept of disability and the discourses used when talking about disability. There is at the present time, no universal understanding of the

concept of 'disability'. The three key defining frameworks will be explored in some detail along with critiques. We will then outline some of the key themes to emerge within disability studies over the last fifty years, encompassing the role of the body within disability, arguments around the impact of genetics on disability, the development of disability politics and recent work looking at disability within a global context. On this last point it is important to note that the vast majority of the work in this area has been carried out in the Western, developed world, and that the history and arguments being presented must be seen in this context. The global context of disability will be covered in the final section of the review.

A Changing Historical Understanding of Disability

Understanding of disability can be traced back to the books of the Old Testament. In ancient Judaism, for example, many diseases and impairments were seen as signs of wrong doing and thus governed the separation and differential treatment of people (Barnes et al, 1999). Barnes et al highlight the writings of Leviticus, in the Old Testament, who:

> "...catalogues those human impairments which preclude the possessor from participating in religious rituals - crooked nose, sores, missing limb, leprosy and skin diseases, and crushed testicles." (Leviticus 21: 16-20)

Ancient Christianity also saw impairments as punishments for sin although some impairments were seen as worthy of charity and healing. This view linking impairment to morality and judging people according to the type of impairment that they have can be seen throughout history, although views on the origins of disability and the treatment that people with disabilities should receive have changed over time and place.

It was not until the end of the Middle Ages and the degeneration of the church that disability, and particularly mental impairment, took on a wider negative social image. The 'mad' were seen to represent 'ambiguity, menace and mockery, the dizzying unreason of the world, and the feeble ridicule of men' (Foucault 1965: 53). It was in this period, towards the end of the sixteenth century that the first 'great madhouses' were opened in Spain and so there was the beginning of the 'treatment' of people with a whole range of mental and physical disabilities. People with disabilities were housed with other 'socially unacceptable' people such as the poor, criminals, beggars, homeless and the mentally ill. By placing people with disabilities into confinement with other marginal social groups the negative image of disability, and particularly mental disability or mental illness was confirmed. Disability became a marginal, negative concept and the disabled became seen as a threat to society and morality (Porter 1997). This image continued throughout the next two and a half centuries with more disabled people forced into asylums and workhouses with the industrialisation of industry in the eighteenth century and the removal of people with disabilities from the workplace (Barnes et al 1999).

It was not until the end of the eighteenth century that asylums became seen as medical institutions and that the 'treatment' of people with 'mental' disabilities was deemed possible (Hirst & Woolley. 1982). The success of the new medical sciences, which were developing throughout the nineteenth century, in medicalising illness and impairment helped to legitimate the 'treatment' of the disabled and the need to keep them in asylums, segregated from the rest of society so that they could receive this treatment. Although the new asylums were an improvement on the old madhouses with the removal of chains and cages, these were replaced with surveillance and discipline. Medicine and surveillance were used to classify and regulate disabled people and differentiate between the normal and abnormal, healthy and sick, and sane and insane. (Barnes et al 1999)

The twentieth century saw massive changes in the understanding of disability and the ways in which people with disabilities are treated.

> By the beginning of the twentieth century, the individual approach to disability - which sees its diagnosis and solution in medical knowledge - was securely entrenched. The focus is on bodily 'abnormality', disorder or deficiency, and the way in which this in turn 'causes' some degree of 'disability' or functional limitation." (Barnes et al, 1999, p21)

Medical expertise has been concerned primarily with identifying the physical or intellectual 'abnormality' and prescribing appropriate treatment, and, in many

cases, a person's impairment has become their master status. It is only in the last thirty to forty years that the focus has begun to widen with the politicisation of people with disabilities, taking in aspects of quality of life, self-advocacy and independent living. The political movement within the disability movement is looked at in more detail later in the review. It is important to start, however, with the key arguments around the definition of the concept and experience of disability itself.

Defining Disability

Throughout the last two decades there has been a widescale development of the politics, understanding and study of disability issues. Much of the work to date has focused on definitions of disability and the problems with definitions currently in use. As touched on in the introduction, the term disability has been defined in a variety of ways over the past quarter of a century alone. The importance of developing an adequate definition of disability is highlighted by Oliver (1990) who suggests four reasons why such a definition is important. Firstly he suggests that human beings give meaning to the objects that they encounter within the social world and orientate their behaviour towards that object according to the meanings they have given it. Therefore, if disability is given a negative, tragic meaning, then disability will be treated as a negative tragedy. His second point is that the definitions attached to disability define who is and is not able to work, thus offering a legitimate social status to people who are not able to work as opposed to people who choose not to work. Oliver's third point is that, in order to reclaim disability as a positive state, the

terms surrounding disability, and their meanings need to be reclaimed and redefined in a positive way. He likens this to the fundamental redefinition work that has been undertaken by women's movements, black movements and gay and lesbian movements since the 1960s. His final point is that the majority of the work that has been carried out in this area has focused on the financial implications of providing care for disabled people. This has led to a lack of studies looking at what disability actually means to disabled people themselves.

The polarity of the two main positions, the International Classification of Impairment, Disability and Handicap' (WHO 1981) and the 'Social Model of Disability' (UPIAS 1976) were outlined in the introduction, along with the most recent attempt to bridge the gap between these two extremes and create a definition which incorporates both the physiological aspects of an impaired body and the social aspects of an inadequate environment. It is useful to reiterate the key points of the definitions in order to look at some of the criticisms of all three models in more detail.

In 1981 the World Health Organisation adopted the 'International Classification of Impairment, Disability and Handicap' (ICIDH), which became the most commonly used definition of disability amongst medical practitioners and other professionals, including medical sociologists. ICIDH states that:

> **Impairment** - an impairment is any loss or abnormality of psychological, physiological or anatomical structure or function.

Disability - a disability is a restriction or lack (resulting from an impairment) of ability to perform an activity in a manner or within the range considered normal for a human being.

Handicap - a handicap is a disadvantage for a given individual, resulting from an impairment or a disability, that limits or prevents the fulfilment of a role that is normal (depending on age, sex and social and cultural factors) for that individual. (WHO 1981)

Although this is still the most commonly used definition of disability, the ICIDH has been widely criticised for placing the emphasis on the individual and not on society. Amongst a number of criticisms it has been pointed out that the ICIDH places the emphasis on the Impairment as the cause of disability and handicap. The environment is seen as neutral and the onus is placed on the individual, with the use of medical aids, to fit in to the environment (Oliver 1990). Rather than being viewed as the result of an inadequate social environment, as may originally have been intended, a number of researchers have suggested that the 'handicap' category gives the impression that handicaps are "merely complex disabilities" (Bickenbach et al 1999). A view echoed by Grimsby et al (1988) and Orgogozo (1994). Birkenback et al go on to suggest that:

"Although identified as a classification 'of circumstances in which disabled people are likely to find themselves', there is never any reference in the handicap classification to features of the social

> world that create those circumstances. It is a classification of limitations of people's abilities." (1999 p1175)

It is important to note, however, as Williams (1999) points out, that despite the many criticisms that have been levelled at it, the ICIDH definition of disability has been adopted, if unconsciously, by the majority of sociologists working in this field to date.

The social model of disability developed alongside the ICIDH, providing an alternative definition of disability. The clearest version of this definition of disability is as follows:

> **Impairment** - lacking part or all of a limb, or having a defective limb, organ or mechanism of the body.
>
> **Disability** - the disadvantage or restriction of activity caused by a contemporary social organisation which takes no or little account of people who have an impairment (whether it is physical, sensory or intellectual) and thus excludes them from participation in the mainstream of social activities. (adapted from UPIAS, 1976)

This definition rejects the idea that the body, and any impairment it may have, has anything to do with an individual's experience of disablement. It was designed to politicise the disability movement's struggle (Bickenback 1999). Writers such as Oliver (1990) and Swain (1993) suggest that it is social

oppression which prevents individuals from participating fully in society and, thus, that the body is immaterial. In addition it has been suggested that medical sociologists are implicitly or explicitly adding to this social oppression through their conscious or unconscious acceptance of the ICIDH. It has further been suggested that the whole area of disability can only be understood and conceptualised fully by disabled people themselves, thus rendering much work done in the area as fundamentally lacking because of the nature of the people carrying out the research.

A number of criticisms have been levied at the social model of disability surrounding the disassociation of the body from disablement. It has been suggested that by removing the body from the debate around disablement the social model of disability is moving in the opposite direction to the sociology of the body which proclaims to look at the same issues (Williams 1990). In addition it has been suggested that removing the body from the realm of sociology gifts it to medicine unchallenged (Hughs and Patersen 1997). Further criticism surrounds the issue of bringing impairment back in to the debate about disablement, with the idea that this allows the social, as well as the bio-medical to have some command over the body. This has led to the development of an area of disability studies concerned primarily with the disabled body. Bickenback et al (1999) suggest that, although it 'provocative', the Social Model of disability as defined by UPIAS is 'not operationalisable':

> "And in this sense it fails the two aspirations of the social model of disablement – first, to provide a workable model for research, and

second to provide advocates with the hard data they need to convince legislators to pass new laws and change old ones." (1999: 1178).

Problems with the existing definitions of disability led the WHO in 1993 to begin the process of developing a new definition framework for understanding disability, based on a revised ICIDH.

The World Health Organisation has been working – in consultation – to develop an up to date version of the ICIDH, combining aspects of the medical and social models of disability, and providing a tool for uniform measurement of disability across the world. In the last year the 'International Classification of Functioning, Disability and Health' (known as ICIDH2) has been released. ICIDH-2 is a proposed scale composed of measures of body functions and structures, Activity, Participation and Contextual factors, which has been designed to incorporate aspects of the social model of disability, and to include environmental factors. It has also been designed with the aim of utilising neutral terminology and avoiding past stereotypes and negative connotations. ICIDH2 is best described by the WHO Information office as follows:

1. The Body dimension comprises two classifications, one for functions of body systems, and one for the body structure.
2. The Activities dimension covers the complete range of activities performed by an individual.

3. The Participation dimension classifies areas of life in which an individual is involved, has access to, and/or for which there are societal opportunities or barriers. (WHO Press Release no. 19, August 1999)

The three elements of ICIDH-2 are designed to be co-participants in a new definition of the aspects which, when combined, create this concept of disablement. It is important to note that they are designed to be seen as equal participants. Equally important is the assertion that ICIDH-2 was designed as a health classification and cannot be seen as more than a way of classifying the consequences of health conditions. Bickenback et al (1999) define ICIDH-2 as follows:

> "The ICIDH-2 embodies what is now termed the 'biopsychsocial' model, a synthesis of the medical and social approaches to disablement. Each dimension of disablement is conceptualised as an interaction between intrinsic features of the individual and that person's social and physical environment. To ensure that the insights of the social approach to disablement are captured, the draft includes a fourth component, a listing of environmental factors that can be used, in conjunction with the other classifications or separately, to identify the determinants of disablement at the body, person or person-in-context levels of human functioning" (1999 p1183)

ICIDH-2 has only just been released in it's revised form and so there has been not time, yet, for critiques of the final form to emerge. Despite the high praise that it has received so far, it is no secret that the disability movement and many people within the disability field, were unhappy from the outset, at the idea of revising the ICIDH, which they see as fatally flawed. It will be interesting to see, in the next couple of years, the reception that ICIDH-2 gets from the different factions involved in work in the area of disability, and in operationalising the definitions available to them. Defining the concept of disability is only one of the areas in which work has been carried out. Debates around the various definitions have led directly to debates about other aspects such as the role of the body in disability theory.

The Disabled Body

The WHO's 'International Classification of Impairment, Disability and Handicap' (WHO 1981), can be seen to have put the physiological body at the centre of the understanding of disability. This definition of disability has been adopted, if not explicitly, by the majority of sociologists working within the field of medical sociology. At the same time, the emergence of the social model of disability and social constructionist notions of the concept of disability itself, relegated the body, at least within disability studies if not within wider circles, to the sideline, unimportant to the construction and maintenance of disability. It was not until the middle of the 1990s that the tide began to turn. An increasing number of challenges to the social model approach have suggested that 'the body' needs to be brought back in to

disability. This re-emergence of the body, and particularly the rhetoric of the 'impaired body' has serious implications for the social model of disability, where the invisibility of the body can be seen as a major weakness. As Shakespeare Explains:

> "There is a danger, in the stress laid on social and environmental barriers and practices, of ignoring the impact of physical and intellectual limitation and suffering on people's lives." (Shakespeare 1999: 184).

This suggests that the social model of disability needs to be modified in order to re-claim the body from the realm of medicine, thus incorporating bodily aspects into a wider understanding of what it is to be a disabled person.

Archer (1995) suggests that the embodied nature of the human being means that, whilst the human being cannot be reduced to their physiological components, nor can they be separated from them. We are constrained by the nature of our physical body, which directly affects what we are and are not able to do. This theme is echoed by Nettleton (1995) who focuses on the increasing emphasis on the management and reduction, if not irradication, of risk behaviours by individuals seeking to maintain a healthy body through lifestyle, exercise, diet and so forth. In a similar vein, Scott and Morgan (1993) focus on the commodification of the healthy body and body maintenance, and Lupton (1994) highlights the increasing importance being placed on bodies that look good, again causing problems for people with 'abnormal' bodies. Hughes and

Patterson (1997) suggest the need for disabled people to reclaim their bodies and challenge the view of them as faulty and unattractive suggesting that a 'Sociology of Impairment' may go some way to meeting the gap between the medical sociologists approach to the body and the disability theorists approach to the body.

Disability Politics

Political activity has increased dramatically over the last couple of decades with the widescale politicisation of disabled people through a range of organisations and pressure groups. The change in the attitudes of people with disabilities and of the wider population is widely recognised to have started in the 1960's in North America with the politicisation of disabled people leading to the denouncing of the stereotypical view of disabled people and the low expectations that they had for their lives (Dejong 1981). Barnes et al (1999) link the move with the political climate of the time, the changes coinciding with the return of large numbers of disabled soldiers from Vietnam and with the high awareness of civil rights in America in the 1960's. The leading writers at the time came from the 'Independent Living Movement' which fought to change the image of disabled people as dependent and to promote self-help, community living and 'normalisation' (DeJong 1981, Wolfensberger 1983). The 'Independent Living Movement' made links between disabled people and key ideologies such as sovereignty, consumerism and freedom of political and economic expression. Their aim was:

> '...to facilitate the reclamation of disabled people's individual autonomy through opposition to self-serving professional domination and inept beaurocratic administration of welfare'. (Barnes et al 1999: p69)

From this starting point a number of avenues have been available to disabled people wanting to become active politically. At the base level research has been carried out looking at access to voting rights. Fry (1987), for example, showed that, an many cases, particularly for those living in residential homes, access to the right to vote depends on the people running the home and on whether on not they choose to register the people living there. It is not just people living in residential homes who encounter problems however. Lamb and Layzell (1994) document the problems encountered by disabled people living at home, particular those with learning difficulties, being unaware of whether they have been registered to vote or not. Problems with access to voting stations (Enticott et al 1992), applying for the right to a proxy vote (Fry 1987), and the difficulties of obtaining accessible political information (braille e.t.c.) have also been documented (Fry 1987; Enticott et al 1992).

Despite the documentation of problems experienced by disabled people attempting to exercise their political rights, Oliver (1990) highlights the considerable number of organisations available to people, many of which have political agendas. He categorises these into four types depending on their aims and who they are run by:

1. *Partnership/Patronage*

Organisations for disabled people; charitable bodies; provision of services (often in conjunction with statutory agencies); consultative and advisory role for professional agencies – e.g. Royal Association for Disablement and Rehabilitation, Royal National Institute for the Blind, SCOPE, Rehabilitation International.

2. *Economic/Parliamentarian*

Primarily organisations for disabled people; single issue; parliamentary lobbying and research; legalistic bodies – e.g. Disablement Income Group, Disability Alliance, American Foundation for the Blind.

3. *Consumerist/Self-help*

Organisations of disabled people; self-help projects; sometimes campaigning groups, or working in collaboration with local or voluntary agencies – e.g. Spinal Injuries Association, Derbyshire Centre for Integrated Living, Berkeley Centre for Independent Living.

4. *Populist/Activist*

Organisations of disabled people; politically active groups, often antagonistic to partnership approach; primarily activities focused on 'empowerment', personal and/or political; collective action and consciousness-raising – e.g. Union of the Physically Impaired Against Segregation, British Deaf Association, Americans

Disabled for Accessible Public Transport. (Barnes 1999, adapted from Oliver 1990: 117-18)

It would seem, thus, that there are a number of avenues available to disabled people wishing to becoming politically active. There is, however, great variability about the groups which they may or may not choose to join. Some of the politically active groups have more power and influence than others (Barnes 1999). In addition to this, it is difficult for charities to become involved in politics, and, furthermore, the beneficiaries of charities are not allowed to in a position to control the charity, thus limiting the level of participation and control open to disabled people in this context (Williams 1989).

It was not until the last couple of decades that significant moves were made to reorganise disabled organisations so that they represented disabled people rather than 'cared for' them (Drake 1996). In Britain this change was sparked by the development of UPIAS in the 1970s, as an overtly political group of disabled people campaigning for the rights of disabled people (Crewe and Zola 1973). This was followed by a stream of other organisations, the most significant being the BCODP, an umbrella organisation which became the voice of political activism by and for disabled people (Campbell and Oliver 1996). Barnes (1999) suggests that the mainstream political parties have, by and large, failed disabled people, and that they have been left to fend for themselves. This has led to successful campaigns around civil and social rights, and equally, to the development of a political empowerment found in collective

action (Barnes 1999). The fragmentation of groups, debates on who should be included in the disability movement, and infighting amongst different groups representing the same people raise questions, however, about how long the movement will be able to last as a positive force for change. One of the issues which may well cause a split between disabled people with inherited conditions and those with disabilities caused by birth defects or accidents is that of the 'New Genetics'.

Genetic Debates within Disability

Some of the most recent debate within the disability movement concerns the use of genetic intervention in human reproduction, predominantly by means of screening programmes for genetic conditions which cause disability. Much of the argument has centred on the extent to which the 'new genetics' can be said to be eugenic – i.e. a direct attempt to improve the human species by controlled breeding for desired inherited characteristics. There are two opposing narratives at work within this debate. On the one hand there is the argument that genetic interventions herald a new era of reduced human suffering and the first step in eradicating a number of 'genetic' conditions which cause disability and death. This strand of the argument is exemplified by the rhetoric surrounding the 'Human Genome Project', completed in 2001, which has been heralded as "the essence of human life" (Conrad and Gabe 1999: 4). On the other hand there is the argument that genetics is a "totalitarian conspiracy to rid the world of disabled people" (Shakespeare 1999:171). This argument

recognises the threat that genetic intervention poses in the form of a covert attempt at eugenics.

It is this second strain of argument that is most prevalent within the sociological literature on disability, supported by proponents of the social model of disability. Shakespeare (1999), for example, suggests that there is a, predominantly covert, narrative amongst genetic researchers and the medical profession themselves that seeks to rid the world of disability and disabled people, seeing disability as a 'major problem, which should be prevented by almost any means necessary' (p174). He cites the widespread use of a 'narrative of tragedy' surrounding the terrible fate of disabled people living with genetic conditions. He also suggests that there are 'Narratives of optimism' surrounding the use of genetics where proponents of genetic technologies are seen to disregard, seemingly with little thought, the possible problems that such technologies could bring with them. Shakespeare's reservations are shared by others in the area. Green (1994), for example, looks at the problems parents may have accessing adequate genetic counselling when they are called on to make a decision regarding the termination of a genetically abnormal foetus. Following on from this, Allanson et al (1997) documented the lack of provision made by many screening clinics to inform women of negative screen tests for Downs Syndrome. The final argument given in the attack on the 'narrative of optimism', is that, at the present time, no genetic therapy is available to babies diagnosed with genetic conditions, and, thus, to talk of the operationalisation of genetic knowledge in this context is to talk about selective termination (Shakespeare 1999).

Other areas of the new genetics have also been explored. Atkinson et al (1997) for example looked at genetic testing and the implications that it has for disabled people. Similarly Nelkin (1997) has carried out work looking at the implications of genetic information for social control.

The majority literature on the use of genetic technologies for the 'treatment' of disabled people uses highly emotive language and imagery. The 'Disability Awareness in Action' group talk about genetic technologies as a 'search and destroy mission' aimed at disabled people (1997), and geneticists have been likened to 'Nazis' in terms of the results of their work, if not the direct intentions (Hubbard 1997; Bailey 1996). Shakespeare (1999) suggests, however, that emotive arguments such as these do little to forward the debate around the use of genetics, although they may prove popular for people venting their anger at a supposed disregard for the rights of disabled people. He suggests, in agreement with Bailey (1996), that the key issue is one of providing screening where no cure is available except termination. In the words of Bailey:

> "This obscures the fundamental difference between prenatal testing and any other way of preventing illness, namely that the 'treatment' which follows prenatal testing – abortion – 'cures' the condition by eliminating the foetus rather than by stopping the condition occurring in the first place." (1996: 149, in Shakespeare 1999).

It is the need to balance the empowerment of women, having the right to chose screening, termination and so forth, and the degree to which such empowerment is manipulated by a desire to rid the world of disabled people (Lippman 1994). With this in mind, Shakespeare (1999) suggests a need to understand the particular impairments that disabled people are living with. Some are more debilitating than others and may require more drastic treatment. He suggests the need for a better understanding, by the medical and genetic professions, of the particular needs and problems experienced by disabled people with a variety of different conditions, in order to decide the most appropriate course of action as far as genetic technology is concerned.

The Global Context of Disability

As touched on earlier, the vast majority of the work that has been carried out looking at definitions and experiences of disability focus on the experiences of disabled people living in the developed world. This is despite United Nations figures which state that approximately 80% of disabled people in the world can be found in 'developing' countries (in Priestly 2001). Priestly goes on to point out that the definition frameworks utilised in the literature on disability are also based on the experiences of the minority of western disabled people (Priestly 2001). Ghai (2001), for example, looks at the experiences of disabled people in India, where disability is associated, amongst other things, with poverty, both for the individual and their families, and frequently can be seen to be both caused by, and to cause poverty. She emphasises the fact that 60 million of the world's disabled people live in India, and that, of these, 65% to 80% live either

in rural areas or in urban slums where there are very few facilities and survival is the aim (Ghai 2001). Western movements focusing on political power and social equality are a long way off:

> "In a society where pain and suffering are often accepted as karma (fate) and learned helplessness becomes a life trait, consideration of disability as a social issue is a difficult goal. To fight societal exploitation, one has to contend not only with external impediments but also with the experience of chronic internalised oppression." (Ghai 2001: 35)

Other issues such as the impact of war on disabled people (Driedger 1987, Bruun 1995) have been looked at, both in terms of war as a cause of disability, for example, through landmines (Priestly 2001), and also as a positive influence on movements towards civil rights and participation, as observed, for example, as a result of the sweeping political changes in many former Eastern Block countries in the last decade (Brichtova 1998).

Access to education and employment are also highlighted in an increasingly globalised world where modern technologies are increasingly influencing industry and employment. In the West this has led to fights for equality of access to education and employment. Access to basic education, let alone equal access to all levels of education, however, although becoming an increasingly important aspect of life for many disabled children and young people across the world, is still denied to many disabled people in 'developing

countries (Peters 1993), and particular to girls and young women (Shah 1995). In the words of Priestly:

> "...disabled men and women throughout the world continue to be disproportionately unemployed, underemployed and underpaid...resulting in conditions of extreme poverty for many millions of their families." (Priestly 2001: 8)

Poverty, therefore, is still key to the experiences of many disabled people across the world, due both to differential access to education, and also to employment itself (Beresford 1996). These are just some of the issues raised in a growing literature on the global impact of disability. The impact of poverty, in particular, is explored within this body of work. Stone (2001), for example, suggests that disability in the 'majority world' should be seen in terms of a framework of deprivation (adapted from Chambers (1983) incorporating:

- Powerlessness is lack of political influence or leverage, exclusion from community and national decision-making, and denial of opportunities for self-representation.
- Poverty, in the basic material sense of the word, is lack of individual and household resources, assets and income (it is often but not always related to poverty experienced by communities and whole regions).
- Disability is institutional, attitudinal and environmental discrimination by a society (community, culture, state) towards people with perceived impairments.

- Impairment is culturally perceived difference in the body/mind; this might also be extended to ill health and weakness that result from malnutrition and overwork in the majority world.
- Vulnerability is living in a situation whereby any unexpected expense (e.g. on medical treatment, a dowry, a burial) or disaster (poor harvest, loss of livestock to disease, conflict) can tip the balance from survival into extreme deprivation.
- Isolation is about lack of access to support, information, education, healthcare, markets, infrastructure, and so forth. (2001: 52-2).

This model highlights the complexity of the combination of issues facing many disabled people living in the 'majority world'. It is important to note here, however, that, whilst this model may prove useful in understanding some of the issues facing disabled people in the global context, the definition of disability that it adopts is purely a social one, opening the model up to many of the criticisms explored in the section on the literature 'defining disability'.

In Summary

Although clearly not exhaustive, the aim of this chapter was to chart key moments in the historical development of the sociological literature on chronic illness and disability as well as to outline current debates in the area.

A considerable amount of work has been carried out over the last fifty years building a view of chronic illness and what it means to the people that live with it. The work has been developed along a number of theoretical paths, looking, amongst other things, at the relationship between the doctor and the patient, power relations, and the day to day experiences of the individuals with a range of chronic illnesses. The vast majority of the work that has been carried out has examined the experiences of people with the more common chronic illnesses and at the shared patterns and themes that have emerged from this work. The impact of the approach traditionally taken within sociological studies of chronic illness is examined in more detail in chapter four where the five key tenets of a traditional sociological approach are identified and contrasted with the key tenets of the traditional approach to the sociological study of disability.

Similarly, it is since the 1960s that the majority of work has been carried out within the sociology of disability, although the historical developments and changes in the treatment of disabled people and ways of understanding disability can be traced right back to the old testament. The disability movement and political activity has developed along with a body of research looking at identity and meaning, the role of the body, empowerment, and more recently, the new genetics and disability on a global scale. The vast majority of the most recent work has moved away from 'expert led' research to focus on the importance of the voice of disabled people themselves within the disability movement, leading to an approach which focuses predominantly on the active agentic disabled person who plays an active role in the research process. As already mentioned, these debates will be taken up in more detail in chapter

four, where we address issues of method within disability research, and in chapter 7 where the impact of the experiences of families with juvenile Batten disease are explored in the light of current disability debates.

3 The Clinical Manifestations and Management of Juvenile Batten Disease

In order to better understand the social impact of juvenile Batten disease on the families of the children and adults with the condition it is important to have a basic grasp of the disease and it's biomedical implications. Having established the key themes raised in the literature surrounding chronic illness and disability, this chapter gives an overview of the biological aspects of Juvenile Batten Disease itself, and highlights the points at which there are links to the themes raised in the sociological literature. Juvenile Batten disease is a neurodegenerative, genetic condition which causes visual impairment, epilepsy and cognitive and motor degeneration and results in a shortened life expectancy. Details of the clinical manifestations of the disease, genetic inheritance, diagnosis, symptomatology and treatment are covered along with current biological and genetic research strategies and recent developments in care and treatment possibilities.

The genetic, biochemical makeup of every human individual is unique, and in-built genetic defects mean that each person has a predisposition towards developing certain diseases. Adams et al (1997) suggest that as many as 6-8%

of hospitalised children are being treated for diseases caused by single defective gene, and as many as 22-31% have diseases which are thought to be gene-influenced. In addition, research carried out in Germany has suggested that up to half of all blind and visually impaired school children have conditions which are caused by hereditary defects (Lorenz 1996). One such group of hereditary, genetic conditions are metabolic diseases. These result from a defect in one or more of the chemical processes occurring in the human organism and controlling either energy production or growth. Metabolic diseases occur due to an absent or malfunctioning catalyst or enzyme which causes a blockage within the cells resulting in a dirth of (often toxic) chemicals on one side of the blockage and a deficiency on the other. Metabolic diseases are inheritable genetic disorders which are potentially fatal as the chemical imbalances which cause them are often untreatable with current medical knowledge. CLIMB (formerly the Research Trust for Metabolic Diseases in Children) state that so far 1300 of this type of disease have so far been identified (RTMDC 1997). The Neuronal Ceroid–Lipofuscinoses (of which juvenile Batten disease is the juvenile onset form) are one such group of metabolic diseases.

The Neuronal Ceroid Lipofuscinoses

The group of diseases now known as the Neuronal Ceroid Lipofuscinoses (NCLs) were named as such by Zeman and Dyken in 1969. The diseases were first documented, however, in the 19th Century, by Stengel in 1826 and again by the English neurologist Frederick Batten in 1903 and 1914. Whilst the term NCL remains in widespread use in Europe and amongst the

genetic/biochemical/clinical/ morphological fraternity, the generic term 'Batten Disease' is now more commonly used in North America and amongst the families of the children with the diseases (Goebel et al 1999). All of the childhood variants of Batten disease and most of the adult types are inherited as autosomal recessive and, as a whole, the NCLs are the most common neurodegenerative disorder of childhood (Jarvela et al 1996, Goebels et al 1999). Current estimates suggest that there is a prevalence of 1.5 to 2 cases of NCL per million children born within Europe (Goebels et al 1999). Figures based on diagnosis within the UK suggest that there are approximately 10 to 15 new cases of NCL diagnosed each year, and, thus, that there are between 100 and 150 children affected by one or other form of NCL in the UK at any one time (Goebels et al 1999).

Despite the group name 'neuronal' ceroid lipofuscinoses, NCLs are in fact generalised diseases affecting all types of tissue and not just the tissue of the brain. It is the secondary degenerative disease that affects solely the brain tissue and results in the 'neuronal' prefix (Porta ed 1989). NCLs are characterised by "retinal and brain atrophy and the accumulation of fluorescent storage bodies in neurons and a wide variety of other cells"[1]. Initially classified with the gangliodoses as Amaurotic Idiocies, the NCLs were reclassified as lipidoses because of the belief that the storage bodies building up within the cells were lipopigments. The similarities between the storage bodies characteristic of NCLs (as seen under a light microscope) and the Lipopigments Ceroid and Lipofuscin led to the adoption of the name neuronal ceroid lipofuscionses in 1969. The build up of these storage bodies within the tissue

cells causes distinctly shaped ultrastructural appearances (curvilinear-crescent shaped, fingerprint shaped and granual osmophilic shaped) which can be recognised under a light microscope and used to aid in both diagnosis and genetic research (Goebel 1999). It was not until 1986 that research into NCL models in sheep led to a breakthrough in the recognition that the storage material was not a lipid, but was in fact a protein (Palmer et al 1986a,b), a finding that was later confirmed in human NCL through the work of Palmer et al (1990) and Hall et al (1991a,b). This led to the suggestion of a further reclassification of NCLs as proteinoses rather than lipidoses:

> "The major advance of the identification of subunit c of mitochondrial ATP synthase as a major component of the storage material in the ovine form of NCL, and in the late infantile, juvenile and adult forms of human NCL, suggested that the NCLs were not members of the lipidoses but represented proteinoses." (Goebel et al 1999: 1).

At the same time as this biochemical research was being carried out there has been a huge amount of research into the genetics of NCLs. The various NCLs were assigned gene symbols on the assumption that there was one mutation per type. The four main forms of NCL were assigned CLN1, CLN2, CLN3 and CLN4, and in 1989 the first NCL gene, CLN3, was mapped onto chromosome 16p11.2-12.1. (Eiberg et al 1989). Genetic linkage analysis was used to localise the gene responsible to chromosome 16p12, with closest proximity to

loci D16S299 and D16S298 (Mitchison et al 1995, Hofmanet et al 1995, Taschner et al 1995). The current genetic status of the eight most common forms of NCL are listed in table two.

Further research is being carried out with the co-operation of a wide range of research scientists from across Europe and beyond and has resulted in the first scientific monograph dedicated to research on the Neuronal Ceroid Lipofuscinoses, which was published in 1999 (Goebels et al 1999).

Table 2. Current Genetic Status of NCL

Genetic	Chromosome	Gene Product	Clinical Type
CLN1	1p32	Lysosomal palmitoyl protein Thioesterast	Infantile, late infantile and juvenile
CLN2	11p15	Lysosomal pepstatin-insensitive Peptidase	late infantile
CLN3	16p12	Lysosomal transmembrane CLN3 Protein	Juvenile
CLN4	not known	not known	Adult
CLN5	13q31-32	Transmembrane CLN5 protein	Finnish variant late infantile
CLN6	15q21-23	not known	early juvenile/ variant late infantile
CLN7	not known	not known	early juvenile/ variant late infantile
CLN8	not known	not known	Northern epilepsy or EPMR

The identification of the genetic status of a number of the forms of Batten disease, including the juvenile form, raises the first two issue in relation to the disability literature concerning the implications of treating genetic conditions and the role of bio-medicine in giving hope to the families of the children with all varieties of this disease. The issues are examined in detail in the latter half of the thesis.

Variants of NCL

There are a number of different forms of NCL. Although there is little consensus on exactly how many variants there are, it is widely accepted that there are four main forms. In addition, four variant forms have been charted (see table 1) and it has been suggested that there may be as many as 15 a-typical subtypes (Porta ed 1989, Koehler et al 1994, Dyken et al 1995, Adams et al 1997). The symptoms of the four main forms of NCL are broadly similar although there are differing ages at onset and the speed of the progression of the disease varies with type.

The four main forms are:

Infantile (INCL or CLN1) – symptom onset occurs between 6 months and 2 years. The disease progresses rapidly with seizures, dementia, blindness and a severe loss of neurones. Death normally occurs in mid childhood.

Late Infantile (LINCL or CLN2) – symptom onset occurs between 2 and 4 years. Symptoms include seizures, blindness, loss of muscle co-ordination,

mental deterioration and dementia. Death normally occurs between the ages of 8 and 12.

Juvenile (JNCL or CLN3) - symptom onset occurs between 5 and 9 years. JNCL is characterised, amongst other things, by loss of vision, seizures, cognitive degeneration, motor degeneration and dementia. Death may occur any time from late teens to early thirties.

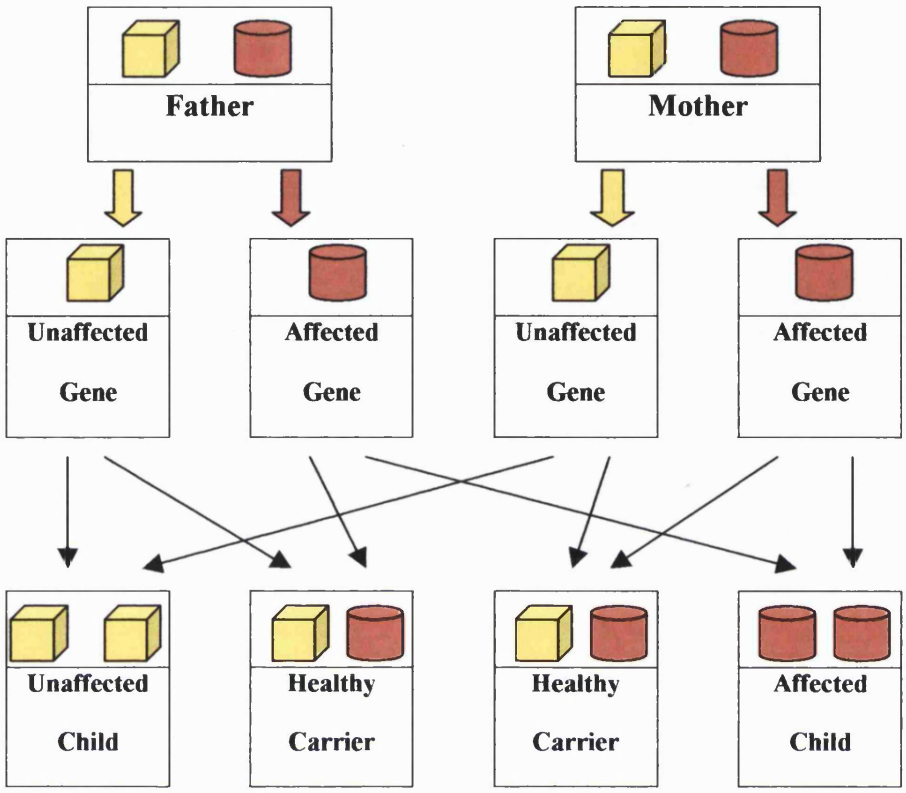
Adult (Kufs or CLN4) – onset is usually before the age of 40 and the symptoms are less severe than in the other forms of the disease. Age of death is variable but life expectancy is shortened. This form of the disease may be inherited through the dominant mode rather than the recessive mode common to the other forms of NCL.

Other variant forms of NCL include the Finnish late variant, early juvenile, variant late infantile, adult recessive, adult dominant and Northern epilepsy forms of the disease. (Scambler 1997). All of the childhood forms of the disease are inherited through autosomal recessive inheritance.

Autosomal Recessive Inheritance

In order to pass a disease on through autosomal recessive inheritance both of the parents must have at least one copy of the faulty gene. It is only when a child receives two copies of the faulty gene, one from each parent, that they are affected by the disease.

Table 3. Autosomal Recessive Inheritance



All of the childhood variants of NCL are such that, if the child reaches adulthood, the symptoms of the disease are of a severity that they are not likely to have children. This means that parents of children with NCL are healthy carriers who carry one copy of the faulty gene each and, thus, are carriers not directly affected by the condition. Consequently the parents are most probably unaware of the fact that they are carriers and that there is a 1 in 4 chance that any child that they have will have the disease, and a 50% chance that they will be a healthy carrier of the condition. Autosomal recessive inheritance is illustrated in table three.

The issue of carrier testing and the implications of genetic conditions on the entire families of people with Batten disease ties in with the arguments within the disability literature surrounding genetics, the development of therapies which aim for eradication of diseases of this type, and the pressure on healthy carriers to decide whether or not to test foetuses or babies for the disease and what to do if the tests come out positive.

Juvenile Batten Disease

Juvenile Batten disease, also known as Spielmeyer-Sjorgren disease (Philippart et al 1994), Spielmeyer-Voigt disease (Hoppen et al 1992, Lorenz 1996), Spielmeyer-Voigt-Sjogren disease (Mitchison et al 1995, Mole 1996) or Stengel-Batten-Spielmeyer-Voigt disease (Koppang 1988) is the juvenile onset for of Neuronal Ceroid Lipofuscinosis. Commonly known as juvenile Batten disease, it is the most common neurodegenerative disorder of childhood. Juvenile Batten disease was originally thought to have an estimated incidence of approximately 1 in 12 500 to 1 in 25 000 births (Porta 1989), however, it is now thought that there are between 50 and 100 children in the UK affected by juvenile Batten disease at any one time. There have been no epidemiological studies carried out in the UK looking at the prevalence of the disease, however, and so prevalence is estimated through the number of diagnoses recorded at various large hospitals, and charity databases (Goebels et al. 1999). This means that prevalence figures in the United Kingdom are likely to be an underestimate due to misdiagnoses and the number of families who, through a variety of reasons, are not represented on charity databases.

The rarity of the disease itself raises interesting issues when confronted with a chronic illness literature developed almost entirely from the experiences of people with more common forms of chronic illness. The rarity of the disease also causes methodological problems which are addressed in chapter four.

Clinical Manifestation

There is little consensus on the age of symptom onset for children with juvenile Batten disease with ranges given from 4 to 12 years (Porta 1989, Rowland 1995, Nardocci et al. 1995, Nelson et al. 1991). However, the average age of symptom onset appears to be at about the age of 6 with diagnosis at the age of 8 (Goebels et al. 1999). Visual failure is usually the first symptom to emerge followed rapidly by increased clumsiness and problems with schoolwork such as speech difficulties, a decreasing attention span and deteriorating cognitive skills. These symptoms are compounded by the increasing visual problems and the degeneration of the short term memory. The most common clinical development of the disease can be seen to have five stages as elucidated in 'Principles of Neurology' (Adams et al 1997):

1. Visual impairment, sometimes preceding retinal changes by months.
2. After approximately two years, the onset of generalised seizures and myoclonus, often with irritability, poor control of emotions, and stuttering, jerky speech.
3. Gradual intellectual deterioration (poor memory, reduced mental activity, inattentiveness). By this stage the movements have usually

become slow and tremulous...to which are added elements of cerebellar ataxia and intention tremor...

4. Stage of severe dementia in which the patient needs assistance to get about, no longer speaks, and may scream when disturbed or forced to move. The muscles are wasted, though the tendon reflexes are lively and the plantar reflexes are extensor.
5. Finally the patient lies curled up in bed, blind and speechless, with strong extensor plantar reflexes, occasionally adopting dystonic postures. Mercifully the illness ends in 10 to 15 years.

Although this is still the most common general development of the disease, it is important to note that treatment and care is changing with a focus on keeping the child/adult as active as possible for as long as possible. Changes in treatment, accommodation and the philosophy of care have led to changes in the way in which the last two stages present with the young adults being kept active and being facilitated in joining community activities (at whatever level is possible) right up to the final stage of the disease. Life expectancy is also extending with children living as long as 25 years after symptom onset.

The key symptoms of juvenile Batten disease manifest themselves in a number specific areas as highlighted by Hall (1987) and Hofman et al. (1999) amongst others:

Eyesight – deterioration of sight is one of the first symptoms apparent within the disease. Short-sightedness leads to uncertain patterns of movement and

problems with judging distances. In the majority of cases blindness follows within 2-4 years of the initial deterioration in vision leaving the child either with some light perception or totally blind. Macular degeneration and retinitis pigmentosa are the most common eye conditions to develop, and other common ophthalmologic aspects of the disease include thinning of the retinal arterioles, typical bulls eye maculopathy and optic disc pallor. Cataracts may also develop towards the latter stages of the disease.

Motor Degeneration - Deteriorating muscle control leads to abnormal patterns of movement. Problems are encountered when initiating or changing movements and movements take on a stuttering, jerky appearance as the ability to control muscle movement decreases. Involuntary, uncontrolled movements may also occur, often aggravated by activity. Eventually retarded motorial activity leads to muscle stiffness and atrophy limiting the movement of the joints. The child/adult moves from walking unaided, to walking with help, to wheelchair use and finally to spending the majority of time in bed. Chewing and swallowing also become difficult as the disease progresses and may become impossible towards the end, resulting in the need for alternative feeding techniques. Control of the bladder and sphincter are also lost leading to incontinence.

Speech Deterioration – Control of both the speech motor system and facial expression declines. "The speech pattern is peculiar with rapid, monotonous, staccato-like repetitive utterances." (Goebels et al 1999; 55). Both verbal and non-verbal communication become increasingly difficult.

Epilepsy – Seizures begin at approximately 11 years, often preceded by minor lapses in consciousness. At the beginning there are an average of four severe seizures per year, but the frequency and severity of the seizures increase as the disease progresses. Progress is not straightforward, however, and symptoms may fluctuate with periods of breakdown which last for approximately a fortnight at a time, resulting in increased epileptic activity and causing anxiety, restlessness, disorientation, delusions, hallucinations and nausea.

Cognitive Degeneration – Problems with cognition develop gradually and can be detected through deteriorating performance at school and reduced attention span and concentration levels. In the early stages it may be difficult to learn new skills and information, memory function and the ability to store and retrieve information will eventually be effected. The ability to sort knowledge and find appropriate information also deteriorates. Cognitive problems can lead to irrationality, incoherence and the loss of awareness of reality. Rapid deterioration may lead to irritability. There may also be aphasic problems and a decreasing ability to communicate, with slower, unclear speech, stammering and a reduced vocabulary – compounded by the deterioration of the speech motor system. The children/ adults are able to comprehend situations and sense moods but are unable to respond to them as their communication skills deteriorate.

Dementia – Several years after the onset of visual problems dementia begins to develop. Dementia results in a deterioration of, once 'normal', cognitive ability (unlike learning disabilities of difficulties where development does occur, albeit at a slower rate) and is gradual and irreversible. It leads to a deterioration in learning ability, social skills and emotional development. Memories are still present although they are often vague, and children with juvenile Batten disease do have some awareness of the fact that they are growing in size and age despite the dementia.

Psychological and Behavioural Problems – Changes in personality are observed as the disease progresses. There is increased sensitivity to failure as the children know what is 'normal' and what is expected of them but are often unable to make their bodies act as they want them to. There is also frustration as the children are growing up physically and emotionally whilst having to contend with deteriorating motor and cognitive functioning. Deteriorating abilities can also lead to irritability, confusion and fluctuating mood changes. Failing eyesight, epileptic fits, physical deterioration and difficulty communicating can all lead to displays of aggression towards parents, carers, teachers and anyone else that is present. Fear of death and dying are also commonly experienced.

Hallucinations – Hallucinations are frequent and may be extremely upsetting and frightening – although they are not always unpleasant (Hofman 1990). Hallucinations are easy to identify whilst the child still has verbal

communication skills but are increasingly more difficult to recognise as verbal communication deteriorates.

Other clinical manifestations include changes in body weight - although body weight is normal at first, there is sometimes a tendency to obesity and later in the disease emaciation occurs – and muscle atrophy, leading to cachexia (muscle and body wasting often seen in the final stages of terminal illness). In addition to this the skin becomes increasingly oily and there may be an increase in hair growth and disease of the hypothalamus and pituitary glands although menstruation may be irregular or violent.

The symptoms of juvenile Batten disease appear to conform to a general pattern with some a-typical variations, although views on the exact progression and causes of each stage are disputed (one such dispute being over whether the onset of epileptic seizures results in degeneration or is the result of degeneration). The speed of the inevitable degeneration, however is variable and uneven. There are periods of stability, periods of rapid deterioration, periods of recovery (although not to the level previously experienced), and periods of gradual deterioration. All of this, when coupled with the continued physical and psychological development of the child and the deterioration of cognitive and motor functioning, can be confusing and frightening at the least and often leads to severe emotional disturbance.

The clinical manifestations of juvenile Batten disease raise a whole range of issues relevant to the chronic illness and disability literatures. The first issue is

that of cognitive degeneration and how that impacts on a disability theory which is built around people who are cognitively 'normal' but happen to have physical differences. The second issue surrounds the whole fact of degenerative disease, compounded in juvenile Batten disease where it occurs over a relatively short period of time, but common to many chronic illness which cause disability. There is also the issue of maintaining an unchanging sense of self identity despite constantly readjusting to new realities, a concept described in the chronic illness literature as 'biographical disruption' (Bury 1998). This raises questions about the possibility of maintaining an unchanging sense of self when faced with both physical and severe cognitive degeneration. The fourth issues raised in this section concerns the profound multiple disabilities caused by the disease and the impact that these have on a disability theory based upon people with simple, singular physical abnormalities.

Diagnosis

An early diagnosis of juvenile Batten disease is crucial due to the inherited nature of the disease as there is a 25% chance that each subsequent child will develop the disease and, due to the rarity of the condition, testing is only carried out as a matter of course where siblings have already been diagnosed. An accurate clinical diagnosis is possible and is relatively straightforward providing that the clinician has a suspicion that juvenile Batten disease is a possible diagnosis. Molecular genetic analysis of lymphocyte DNA can be carried out to establish whether there is the deletion in gene CLN3 on

chromosome 16 which is typical of juvenile Batten disease (Goebels et al. 1999). Diagnosis may be missed or delayed however, until the child has had symptoms for a number of months or even years and has undergone a range of tests because of the slow progress of the disease and its similarity to other NCLs and neurological diseases (Porta 1989). The time that diagnosis takes is speeding up as awareness of the condition grows. In Holland, for example, ophthalmologists are consulted as a matter of course and information from parents and teachers on the child's behaviour is also used to aid diagnosis (Hall 1987).

The development of visual problems is often the earliest sign of the disease and so it is frequently the child's optician or ophthalmologist who first notices that something is wrong. The ophthalmologist can detect the loss of cells within the eye which is symptomatic of the various childhood forms of the disease. This, however, is merely the beginning if the process because this type of cell loss is typical in a number of more common diseases of the eye. If Batten disease is expected then the child will be sent to a neurologist who will be able to carry out more extensive testing. Various diagnostic tests can be carried out to confirm a diagnosis of juvenile Batten disease.

The most common of these are:

DNA Testing – Molecular genetic analysis of lymphocyte DNA can be carried out to establish whether there is the deletion in gene CLN3 on chromosome 16 which is typical of JNCL.

Blood Tests – a blood sample is taken and the white cells are examined for holes or vacuoles common to JNCL.

Eye Examinations – Abnormal dark patches can be found on the back of the eye in some forms of NCL.

Urine Tests – unusually high levels of the chemical dolichol may be found in the urine of people with NCL.

Tissue or Skin Sampling – rectal, conjunctival or skin biopsies may be carried out. A small piece of skin is examined under an electron microscope and abnormal patterns of storage material can be identified. The patterns are described as 'curvilinear bodies', 'fingerprints' or granular osmophillic deposits', and the type of pattern identified indicates the type of NCL that a child has.

Computed Tomography (CT) or Magnetic Resonance Imaging (IMR) Scans – These scans produce images of the brain. The images can then be examined to reveal areas of the brain that are decaying which is a sign of NCL.

Electoencephalogram (EEG) – This is a brain wave test. Patches are placed on the scalp to record electrical currents inside the brain. Certain patterns suggest that a patient has seizures which is another indication of the disease.

Visual Evoked Potentials (VEP), Somatosensory Evoked Potentials (SEP) and Electroretinogram (ERG) – These are all electrical tests which use brain waves to identify eye conditions common to NCLs (Batten Disease Research and Support Association 1997).

The skin biopsy can also be used as a prenatal test for juvenile Batten disease, however, the rarity of the disease means that it is not used as a matter of course and is only available routinely where siblings have already been diagnosed with the disease.

The wide range of tests available in the diagnosis of children with juvenile Batten disease raises the issue of the role of diagnosis in the development of the disease. Within the chronic illness literature diagnosis is seen as a momentous occasion resulting in the labelling of an individual as chronically ill and the possible change in their master status identity. With juvenile Batten disease the diagnosis, while clearly momentous in itself, and often long awaited, may come second to the severity of the symptoms already being experienced.

Medical Management

Once a diagnosis of juvenile Batten disease has been made, doctors are able to actively manage the condition. There is no known treatment at the present time that can either stop the progression of, or reverse the effects of juvenile Batten disease, however, over the last twenty years there has been a growing interest in research in this area with the aim of finding a cure or, at the very least, treatments which can slow down the progression of the disease. Although no general treatment is available, symptomatic and supportive treatments are used and medication can help to control symptoms. A wide variety of practioners – occupational therapists; physiotherapists; speech therapists; rehabilitation workers; ophthalmologists and counsellors, amongst others - are involved in

the medical management of the condition and the care of the child and their family. Anti-convulsants are used to control the epilepsy, anti-hallucinogenic drugs control the hallucinations, sheepskins and air matresses prevent bedsores caused by the ataxia and nasogastric tubes or pegs are used for feeding when the swallowing reflex is lost. The two key problems that tend to require hospitalisation are the uncontrolled epilepsy (sometimes resulting in 'status epilepticus') and the possible need to insert a peg for feeding when the swallowing reflex is no longer working. Most of the other symptoms can be treated in the home as long as the medication levels are correct and the parents are given the appropriate training and support.

The emotional and physical needs of the child/young adult also require careful management, particularly in the latter stages of the disease, where the disabilities experienced by the young person are becoming more severe. Shroijen (1994), an educational psychologist working at Bartimeushage in the Netherlands, outlines the key issues requiring medical and non-medical management from this point. In the latter stages of the disease process the young people develop multiple, profound disabilities. They experience mental and physical deterioration, diminishing verbal communication, aphasia, incontinence and gradually find that they are spending more and more time in a wheelchair. At this stage they will need emotional support to cope with the changes that they are experiencing, increasing help with daily living skills, and emotional support to prevent feelings of isolation commonly experienced in coming to terms with all of the changes going on within and around them. From here the young people experience gradual immobility and increasing

physical vulnerability. Verbal communication and the ability to act on their own volition will have eroded to the extent that neither are possible without assistance and carers are increasingly relied upon to interpret the signals and tokens of communication. There is also a danger that apathy will follow if stimulation and interpersonal contact is not maintained, and so both become increasingly important aspects of care. The combination of physical vulnerability and symptom degeneration herald the onset of an increasing need for more intensive nursing care and, eventually, the need for terminal nursing are in the last few weeks of life. Although many of these aspects of care are not, strictly, medical, they can all be encompassed within the auspices of the role of the holistic practitioner in managing the care needs of a person with juvenile Batten disease.

A number of experimental treatments for juvenile Batten disease have been tested over the last 25 years, predominantly exploring the possibilities of using dietary supplements to control the condition. Santavuori et al. (1990), for example, studied the development of 26 Finnish patients who were treated with antioxidants (vitamins B_2, B_6, E and sodium selenite) and compared their development with that of 17 German patients who had not been given the antioxidant drugs (Kohlshutter et al. 1988). Santavuori found that there was a significant difference in the development of intellectual, motor function, language loss in the patients who were treated with the antioxidant drugs suggesting this as a possible treatment. However, Hofman et al. (1999) note that observed differences may be due, at least in part, to the higher overall levels of care available to the Finnish patients. Polyunsaturated fatty acid

supplements have also been used in an attempt to control the symptoms of the disease. Bennett et al. (1994) used polyunsaturated fatty acid supplements to treat 6 patients with juvenile Batten disease who were demonstrated to have abnormally low levels of membrane phospholipids. The treatment was followed for between 4 and 7 years, during which time mental development remained stable for most of the patients and fine motor function and vision remained stable in the two youngest patients. Bennett suggests that polyunsaturated fatty acid supplementation may arrest the progression of juvenile Batten disease if the treatment is started early enough. However, large scale testing of the treatment is needed and has yet to have been attempted. Although research continues, at the present time there is no treatment in widespread used that is documented as making a significant difference to the prospects of people with juvenile Batten disease, and medical management remains concerned with controlling symptoms and providing care through the emotional and social aspects of the disease.

Again, the medical management of the disease raises a number of issues within the literature. There is the issue of normalisation and the fact that this is not possible in a disease with the severity of symptoms that juvenile Batten disease entails. Again we also have the issue of the inescapable role of bio-medicine, previously identified through the importance of the hope provided by the researchers. In addition to this there is the issue of care provision and the specialist care needs of this group which may best be served in a specialised environment away from the general population. And finally there is the need to care for the family of the child as well as the child themselves which ties in

with issues identified within the chronic illness literature around family involvement in the disease processes .

Current Research

Much of the current research that is being carried out on juvenile Batten disease is concerned with elucidating the exact nature of the CLN3 gene. The protein within CLN3 which is dysfunctional was identified as a membrane protein five years ago, but researchers are still trying to discover the exact nature of the protein and it's purpose (Gardiner 2001). It has been found that protein homologues similar to those found in CLN3 exist in both yeast and a worm called C.elegans, and that the worm is particularly useful as a research tool because it has only 1000 cells (in comparison to the many millions that make up a human being) and because all of its 19 000 genes have been mapped (Gardiner 2001). In addition work has been carried out at UCL to develop a "knockout" mouse model of juvenile Batten disease where the CLN3 gene does not function, allowing the possibility of more research into the exact nature of the CLN3 membrane protein (Mitchison et al. 1998). Lane et al. (1996) have also been looking at the nature of the neurodegeneration that occurs in NCLs and have established that apoptosis (or cell suicide) is the mechanism through which the neurodegeneration occurs. Apoptotic neuronal cells were detected in brain samples of patients and also in a canine model and in brain and retinal samples from an ovine model of NCL. This is the first neurodegenerative disease involving the brain and retina where apoptosis has been confirmed as

the mechanism of photoceptor and neuronal cell death in both humans and animals.

Further research is targeting the diagnostic techniques currently in use to diagnose juvenile Batten disease with the aim of simplifying them and making the more accurate. Taschner et al. (1995) have been carrying out research into haplotype analysis as a way of making carrier detection and prenatal diagnosis more accurate. Autti et al. (1996), alternatively, have been looking in to the use of magnetic resonance imaging (MRI) to achieve more accurate diagnosis. They studied 30 patients with JNCL and 43, age matched, health volunteers, and found that the MRI results correlated significantly with decreased intelligence, motor problems and speech disturbance. Autti et al. Have suggested that the absence of pathological MRI results in the early stages of the disease may help to assess the severity of prognosis in young patients. Two further branches of research were reported at the International Neuronal Ceroid Lipofucinoses Conference held in Oxford in 2000. Here it was reported that a new enzyme-based diagnostic tool is being developed for use with NCLs and also that 'gene chips' or 'micro arrays' are being developed, whereby thousands of genes can be viewed on a glass slide, allowing tests to be carried out on the functionality of activity rates of particular genes, with the potential of exploring the disease process of a whole range of genetic diseases (Gardiner 2001).

The third key area in which research is currently being carried out relates to the development of treatments, potential therapies and the clinical management of the disease. As already highlighted, a number of treatments have been

explored using dietary supplements as a way of slowing down or arresting the progress of the disease. The majority of work, however, is focusing on gene therapy and cell stem transplants. Stems or progenitor cells are cells which have the ability to transform themselves into any other type of cell and work is currently being carried out to see whether it would be possible for these stem cells to transform themselves into brain cells and replace then brain cells that are killed off during the disease process (Gardiner 2001). Professor Gardiner also reports on the possibility of developing gene therapy whereby the faulty gene, as identified, can be replaced by a new healthy copy of the gene. At the present this is the best hope for curing juvenile Batten disease, although, clearly, any such gene therapy is still a long way off.

Very little research has been carried out looking at the social implications of juvenile Batten disease on the child and their family. Labbe (1996) carried out research in Australia looking at the family functioning of care givers and the emotional states of primary care givers of children with Batten disease in comparison with the emotional states of primary care givers of chronically ill children with less severe illnesses. Primary care givers of children with Batten disease were found to be significantly more anxious and depressed, reported greater negative effects on their schedule and health, and perceived their families as less cohesive. The remaining social research that has been carried out looks more specifically at the social care needs of children and young adults with juvenile Batten disease. Coen de Jong (1996) identifies six key areas of care, incorporating the accumulation and interpretation of information by the family; the impact of the emotional development of the child on their ability to

cope; the need to make knowledge available to those involved in the day to day care of the child; and the need to provide emotional support for the families and carers. Shroigen (1993) has carried out research looking at the psychological impact of the disease on the children and young adults, and Von der Dunk and De Jong (1992) have identified the need to support carers through the terminal stages of the illness as well as the family and the child themselves. Scambler (1999) looked particularly at the social care needs of young adults with juvenile Batten disease and found, amongst other things, that the majority of parents are not happy with either the way they are given information, or the amount of information they were initially given about the disease and that carers, clinicians and therapists need to work in partnership with the family in order to provide the most appropriate and effective care.

The need for research and the involvement of families in research with the hope of finding future cures and treatments links with the issues previously identified around the specific implications if a rare condition, the role of bio-medicine, and the impact on the families, in that families are tied in to multiple research projects which provide their only hope for the future.

Summary

The relevance of juvenile Batten disease in providing an exemplar of the potential problems of and changing nature of chronic illness and disability can be seen in the issues raised, through the clinical manifestations of the disease, for the current literatures surrounding chronic illness and disability. The nature

of juvenile Batten disease as a chronic, terminal disease causing profound, multiple disabilities should ensure its inclusion in a sociological analysis of these areas and provides interesting challenges for our existing theoretical understanding.

Table 4 (on the following page) highlights the issues to be explored within this thesis, as identified outlined in the introduction, as related to the key biomedical aspects of juvenile Batten disease identified in this chapter.

Table Four

BIOMEDICAL FACT OF JUVENILE BATTEN DISEASE	ISSUE RAISED
Genetic Disease	Gene Replacement Therapy, The New Genetics, Impact on the Family
Very Rare Condition	Very Limited Literature, Small Sample Population, Methodological Difficulties
Diagnosis is difficult	Rarity, Diagnostic Techniques Available,
Degenerative Disease	Constantly Changing Reality over a Relatively Short Period of Time
Profound Multiple Disabilities	Severe Disability, Affecting Many Aspects of the Individual
Severe Cognitive Degeneration	Gradual Loss of Skills, Focus on Maintenance not Learning New Skills
Short Term Memory and Communication Skill Losses	Loss of Means of Maintaining Sense of Self
Bio-medical Care is Essential	Bio-medicine Gives Only Hope for Present and Future
Specialist Care Needed	Normalisation is not Possible
Family Role is Crucial	Carriers, Carers, Proxy Information, Live with the Disease, Continuity

4 Methodology

Overview

A variety of, predominantly qualitative, methods have been used by researchers attempting a sociological analysis of the impact of chronic illness and disability on society, the most common being interpretativist; interactionist; symbolic interactionist; and ethnographic approaches. This study starts from a critical realist ontology, as explored in chapter one, allowing the opportunity of challenging the postmodern, social constructionist approach to chronic illness and disability particularly found within the literature on disability theory (Oliver 1990, Swain et al 1993). In this chapter we explore the methodological approaches to the sociology of chronic illness and disability that have traditionally been adopted before focusing on the use of a critical realist inspired ethnographic approach as a tool for understanding the experiences of chronic illness and disability from the constructed understanding of families with juvenile Batten disease, relocating their understanding and experiences within a critical realist ontology. From here we look at the particular methodological issues posed by the study of juvenile Batten disease itself and outline the methods of data collection and techniques of data analysis used within this study.

Locating the Research Within a Critical Realist Ontology

Critical Realism, as examined in Chapter One, is an ontological position developed, predominantly through the work of Bhaskar, which incorporates an argument for the reality of the social world, a three tier classification of the world and a threefold classification of the basic ontological properties that are necessary to the existence of a social reality, and thus to the practice of social science itself. Each of these aspects need to be taken in turn in order to understand the theoretical basis for this research and what it adds to our exploration of sociology's understanding of chronic illness and disability.

At the heart of Bhaskar's theory of critical realism is the dismissal of the 'epistemological fallacy'. This, he suggests, is the conflating of the epistemological and the ontological, the reduction of 'what is' to 'what we know about'. The adoption of a critical realist perspective allows us to look at society at the level of individual understanding and action with the knowledge that these are merely a part of what constitutes society, and that, whilst individual action may influence the nature of social structures, society also exists independently of human action. Realism looks beyond the individual, suggesting that society is not simply a grouping of individuals sharing meanings and understandings which can be interpreted as social structures. Rather, social structures are seen as 'real' structures which are, at least in part, existent as entities separate from individuals and their understandings and meanings (Silverman 1985). In the words of Bhaskar:

> ...people do not create society: for it always pre-exists them and is a necessary condition for their activity. Rather, society must be regarded as an ensemble of structures, practices and conventions which individuals reproduce or transform but which would not exist unless they did so. Society does not exist independently of human activity (the error of reification) but it is not the product of it (the error of voluntarism). (1979: 45-6)

Having established the existence of the social world as a 'reality', Bhaskar went on to suggest a three tiered classification of the world and what lies within it. As highlighted in the introduction, he refers to this as the three dimensions of the 'real', the 'actual', and the 'empirical' (Bhaskar 1975). The real can be seen to have two aspects to it. Firstly it refers to anything that exists, whether we have knowledge of it or not, and regardless of whether or not it is observable as an object. Secondly, it refers to the structures and power that lies within the objects:

> 'Whether they be physical, like minerals, or social, like bureaucracies, , they have certain structures and causal powers, that is, capacities to behave in particular ways, and causal liabilities or passive powers, that is, specific susceptibilities to certain kinds of change.' (Sayer 2000: 11)

Thus, sociologists need to look not just at what necessarily happens but also at the potential or possibility of other things happening due to different

combinations of real objects and their structures and powers. The domain of the actual then carries both events and experiences and can be seen to contain the results of the activation of the power, or generative mechanisms, contained within the 'real'. The actual may or may not be observable but the events and experiences that it contains are beyond those of the empirical. The third level of the world, from the perspective of critical realism, is the domain of the empirical which contains observable experiences, which we are able to observe regardless of whether we have knowledge of the 'actual' or 'real' aspects of the world that the empirical is contingent to (Sayer 2000).

Having established his three domains, Bhaskar (1989) goes on to suggest that "Only on the metaphysical assumption that some things are necessary and, at least relatively, enduring can we reasonably set out to practice science or study society" (Scambler & Higgs 2000 p??). He goes on to establish three basic, ontological premises regarding the nature of social reality, intransitivity, transfactuality and stratification.

Intransitivity	the existence of entities which are independent of their identification. There are things which can exit independently of our knowledge of them.
Transfactuality	the activities of these mechanisms are continuous and invariant, stemming from their relatively enduring properties and powers, they are also morphogenetic, having the ability to change their shape of form.

Stratification Understanding social reality is not reliant on surface data gained through horizontal study of the three domains. Rather, experiences and observations must be explained vertically in terms of the generative mechanisms indispensable for their realisation. (Scambler & Higgs 2000)

Bhaskar's 'critical realism' incorporates 'tranformational' and 'relational' models of social activity and society, which enable us to understand and study the social world (Scambler & Higgs 1999). His transformational model of social action suggests that social action and human agency exist interdependently and essentially:

> society is both an ever-present condition (material cause) and the continually reproduced outcome of human agency. And praxis is both work , that is, conscious production, and (normally unconscious) reproduction of the conditions of production, that is society. (Bhaskar 1989: 35)

Bhaskar refers to this, respectively, as the duality of structure and the duality of praxis. When addressing the social world, he states that people do not create society as it always pre-exists them, and yet at the same time, society could not exist except through their existence.

This brief outline of the key tennets of critical realist theory highlights the theoretical foundations upon which this research is based. Having established what the theory says, it is necessary to look at why it is useful in this particular study and what it adds to our understanding of existing theories surrounding chronic illness and disability and their potential problems.

Critical Realism and the Chronic Illness and Disability Debate

Sayer (2000) suggests three reasons for adopting a realist approach when attempting to understand the social world and particular aspects of it. He suggests that it provides a useful alternative to existing methodological and philosophical positions. Firstly he suggests that critical realisms provides us with a middle ground between the positivistic, empirical approach and the relativistic approach to scientific study. Following on from this, critical realism can be seen to provide an alternative approach to the 'law-finding science of society' which adopts the methodologies and aims of the natural sciences, and the 'interpretivistic reductions of social science' when studying the interpretation of meaning (p3). The third benefit of critical realism is that it provides a middle way between the closures and reductionism that can be found in modernism, and the absence of any closure, regulation or certainty represented by postmodernist approaches to the social sciences. Thus, critical realism allows for the 'openness, contingency and contextually variable character of social change' whilst allowing for the reality of the social itself regardless of our knowledge of it (Sayer 2000: 3). Critical Realism offers us a "challenging alternative to the relativism of constructionism itself" (Williams

1999: 805). It further allows us to recognise the "role of 'meaning' in social life without accepting that this dissolved the constraining power of social structures" (Silverman 1985: 29) and to "recognise the reality of social relations while understanding that they could not exist independently of human activity" (Silverman 1985: 35)

In relation to the chronic illness and disability debate, the critical realist perspective allows us to look beyond the experiences of the individuals towards the causal factors or generative mechanisms which shape those experiences. It provides a way of challenging an approach which looks only at the individuals day to day experiences, remaining at the level of the empirical. The same can be said of the disability literature which has a very strong focus on the experiences of disabled people themselves, to the extent that it has been suggested that non-disabled people should not study disability because they cannot hope to understand what it actually means to live as a disabled person (Oliver 1990). So, having established the theoretical basis for the study we need to establish the methodological approaches traditionally used within the sociology of chronic illness and disability and to identify the reasoning behind the choice of method being used in this study.

Traditional Methodological Approaches to the Sociology of Chronic Illness and Disability

There are five key themes which can be seen to illustrate the traditional methodological approaches to the sociology of chronic illness and a further five

which are particular to the sociological debates around disability. By identifying these we are able to identify possible problems with adopting traditional approaches when looking at a condition such as juvenile Batten disease. These themes are also represented in tabular form later on in the chapter.

The first theme is that the sociology of chronic illness, as was illustrated in the literature review, can be seen to have developed as a challenge to the traditional, bio-medical, focus of studies looking at aspects of health and illness. Sociologists in this area have suggested that the nature of chronic illness itself, and the fact that it is rarely curable, reduces the importance of bio-medicine when studying the lives of people with this group of conditions. They suggest that it is the experiences and difficulties faced when living on a day to day basis with a chronic illness which characterise this group of illnesses as different from others, and, thus, that it is on these day to day experiences that we should be focusing. The second theme, therefore, is that traditionally sociological research within this field has been qualitative research which focuses on the experiences of the individuals with the diseases and, to a lesser extent, on their families. The research can be seen to be user-led and interpretativist, and can be recognised as the approach of many of the studies commonly cited in the literature, such as those of Robinson (1988) on multiple sclerosis, Pinder (1990) on Parkinson's Disease, Anderson (1998) on stroke, and Scambler (1989) on Epilepsy.

The third theme links to the previous one in the sense that user-led research relies on the experiences of the individuals themselves. Although the views and experiences of the family may well be studied in their own right, and there is an acknowledgment that chronic illness is something which effects not just the individual but the whole family, carers are seen very much as carers who only really experiences the disease through the experiences of the individual. This is something that will become increasingly important when we look at the particular methodological problems specific to the study of people with juvenile Batten disease.

Fourthly, the traditional sociological approach sees the body as incidental to the experience of living with a chronic illness. It is the experiential aspects which are key to understanding what it is like to live with these particular conditions. This ties in with the final theme, which states that chronic illness can be seen as a social construct in the sense that the experiences that people have, and the understanding that they develop all hinge on the diagnosis that they receive. Again, a considerable body of research has been carried out which focuses on the importance of the diagnosis from the point of view of stigma (Goffman 1968, Scambler 1989) and the sick role (Parsons 1951). These five themes summarise the approach most commonly adopted in this area of sociological exploration.

Moving on from here, five similar themes can be identified when looking at traditional sociological approaches to the study of disability. Firstly, disability research has also focused on user-led research with the experiences of the

individuals with the disabilities being seen as the primary source of information and understanding. Following on from this, sociological accounts of disability also focus on the use of insider accounts of what it is like to live with a disability. There is a feeling amongst disability theorists that only people who live on a daily basis with a disability are able to fully understand what it is like to live with a disability, and, thus, that only people with disabilities should be allowed to carry out this type of research (Oliver 1990). The third theme suggests that it is the environment which disables people and that the body is, therefore, immaterial, when looking at the experiences of people with disabilities. Again the similarity between this and the traditional approach to the sociology of chronic illness can be noted.

The fourth theme to be identified is that of a challenge to the dominance of bio-medicine. Again, if the body is merely incidental to experiences of disability, then bio-medicine also becomes marginalised and it has been suggested that definitions of disability which hinge on bio-medical judgements about impairments are inappropriate. Leading on from here, the final theme is all about power and agency. Much of the research carried out in this area focuses on the importance of agency and assumes that the disabled person is just as capable of acting in an agentic way as the non-disabled person. The focus therefore is on power relations and the means employed by non-disabled people to 'disable' disabled people. These themes, although only briefly explore here, encapsulate the traditional approaches that have been taken to the study of chronic illness and disability. The need for a different approach when studying a condition such as juvenile Battens disease and when seeking to look

beyond the surface to the generative mechanisms which influence our experiences and understanding are explored in the next sections. Followed by an outline of the problems specifically associated with studying juvenile Batten disease and a table summarising the key points of all three approaches, that traditionally adopted within sociology, that suggested by the specific nature of this condition, and that adopted in this particular study, starting with the approach being adopted within this study.

An Ethnographic Approach

Ethnographic methodology is one way of accessing the experiences of the families and the range of other people who shape the experience of juvenile Batten disease as a whole. Ethnographic research has developed around the basic principles of anthropology, the main elements of which include the concepts of culture, holism and in-depth studies (Sarantakos 1998). Culture is seen as a system of shared beliefs and behaviours, and as an entity in itself which needs to be studied. Ethnography seeks to understand how culture develops, changes, or may be destroyed, and the context in which it exists. It "tries to understand the patterns of behaviour, values, norms and standards as experienced and practiced by people" (Peoples and Bailey 1988). The second element of an ethnographic approach is that of holism, which is described by Sarantakos (1998) as such because "it perceives human action in the context of the whole system, for which it is an expression. Social action is part of the whole and derives a meaning through the place and purpose it has in the context of the system" (Sarantakos 1998: 196). Thus ethnography looks beyond

individual experiences at the society within which human experiences are found, and the social mechanisms which help to shape them. The final aspect of ethnography is that it is interested in looking beyond the surface experiences of its subjects. Ethnography uses in-depth studies to look beyond the surface of the information which they are initially given, to observe the subjects in context and examines the ways in which they experience their experiences as well as to look at what they themselves say about their experiences.

The Criteria of Ethnography

Zahalik and Green (1992) suggest twelve criteria which ethnographic research fulfills. Seven of these criteria are directly related to this research, as detailed below:

- *Social relationships* ...assumes that researchers establish a long-term and diffuse relationship with the respondent...over a long period of time and in a variety of contexts.
- *The researcher as learner*...the researcher assumes a role that is inferior to that of the researched. The researcher knows very little and wants to learn from the respondent, who knows more about the research issue.
- *First-hand information*...essential information gathered by the ethnographer is gained through direct contact with the respondents...

- *Long-term observation*...requires long-term contact with the respondents, that is, long enough to allow experience with regular routines and patterns of life as well as with reactions of people who find themselves in different situations and circumstances.
- *The ethnographer as research instrument.* Practically the ethnographer...is the instrument. The whole personality of the researcher is involved in the research.
- *Eclectic approach* ...the use of different data collection techniques...to enhance 'scope, density and clarity of constructs' gained through one way or another.
- *Holistic approach*...it attempts to understand social structures and processes of elements of the system in terms of the whole socio-cultural system, and in that it assigns meanings to parts of the system by 'connecting' them to the whole. (Zaharlik 1992: 119-21; in Sarantakos 1998)

Criticisms of Ethnography

The main criticism of ethnography is that it favours a culturally realtivist approach to the study of the social world. Ethnography, it has been suggested, developed within the anthropological field as a way of seeking the values, logic and understanding of particular societies or groups from a position of cultural relativity, with said values being seen to have an internal logic of their own.. Anthropologists such as Clifford Geertz (1973) have taken the social constructionist element of ethnographic research to the level of suggesting that

the view of the ethnographer is one view amongst many, none of which are inferior or superior to any other, and adopting a postmodern position of total relativity.

In response to this, however, Abercrombie et al suggest that it is clear that the ethnographer can accept that they are producing a reflexive interpretation of the group that they are observing rather than an objective description without adopting the postmodern position of cultural relativity and abandoning the notion of a wider, objective reality. In this way the ethnographer can observe and interpret the understanding of the subject group whilst acknowledging both their own influence on the description that they are producing and the existence of a wider epistemology such as critical realism (Abercrombie et al 1994).

Why Use Ethnography in This Study?

Having outlined the key tennets of an ethnographic approach, and some of the criteria which need to be fulfilled in order to adopt this approach, all that remains is to identify the reasons why ethnography has been used in this study. The reasons for this choice fall in to two categories, those relating to the theoretical basis of this study, and those relating more specifically to the 'problems' surrounding the study of juvenile Batten disease itself.

Critical Realism provides us with a framework through which we can look beyond the level of the empirical at the actual and the real, to establish not just observable experiences and events but also non-observable experiences and

events and the generative mechanisms which the shape the events. Ethnography, similarly, seeks to look beyond the immediately observable surface experience in an attempt to discover the meanings of experiences in the wider social context within which they occur. In addition to which, ethnography seeks to look not just at the individual experiences but at the social world as a whole. Thus the basic aims of the theory and method concur.

An ethnographic approach also fits in with the particular problems of studying juvenile Batten disease (as expanded on below). An ethnographic approach allows the acknowledgment of the role that the researcher themselves plays in the research process. In addition it advocates seeking information over an extended period of time, from a variety of sources, collecting information both directly and indirectly through interview and observation. The importance of these aspects of ethnography in relation to the study of juvenile Batten disease will become clear in the next section where the specific methodological problems surrounding the study of juvenile Batten disease are identified.

Methodological Issues surrounding the Study of Juvenile Batten's Disease

A number of problems arise when attempting to study the experiences of families with juvenile Batten's disease, some of which are particular to juvenile Batten's disease itself and some of which are shared with a range of other rare, degenerative chronic conditions. The three key areas in which specific issues affecting the choice of study method arise are in the impossibility of carrying

out user-led research, the inability to move away from the expert knowledge base, and the sampling problems caused by an extremely limited sample population.

Insider, user-led accounts are favoured within much of the recent experientially based, interpretativist research that has been carried out within the sociology of chronic illness. Interpretativist studies focus on the ways in which people understand health and illness and the concomitant interactions between the individual and society (Radley, 1994). The social disadvantage accredited to people with a chronic illness and/or disability is seen as resultant of the ways in which the individual interacts with society and vice versa (Williams & Wood, 1988). This kind of approach focuses on the complexities of the subjective experience of chronic illness in everyday life and is the most common type of research carried out in this field. The research consists predominantly of smaller scale, qualitative studies, focusing specifically on the views of the individual and their immediate family.

Herein lies the first problem. There are two unassailable reasons why it is not possible to compile Insider accounts of the subjective, day-to day experiences of living with juvenile Batten's disease. Firstly, the combination of symptoms that occur in this condition make it difficult or impossible to gain complex, meaningful information from older teenagers and young adults. Deteriorating muscle control leads to retarded motorial activity, muscles begin to waste and become atrophied and control of both the speech motor system and facial expression declines, making communication increasingly difficult. In addition,

mental deterioration results in loss of short term memory function, the ability to store and retrieve information and the ability to sort knowledge and find the appropriate information, and can lead to irrationality, incoherence and the loss of a grip on reality. There may also be aphaisic problems and a decreasing ability to communicate, with slower, unclear speech, stammering and a reduced vocabulary. The children will be able to comprehend situations and sense moods but cannot respond to it as their communication skills deteriorate. Concurrently there is the gradual and irreversible onset of pre-senile dementia, further affecting memory functions and awareness of reality. In addition to the problems caused by the specific symptoms of the disease, further complications arise from the fact that none of the parents that I talked to in the course of the research – encompassing approximately half of the families in England with the condition – had told their children what their diagnosis was or of the implications that the diagnosis has for their long and short term futures. This makes direct and indirect questioning of the children and young adults both difficult and unethical. Information on the effects of the condition on the everyday lives of the young person and their families has, therefore, contrary to sociological preference, to be collected solely from proxies or observation, thus requiring an eclectic approach to the collection of data from a wide range of sources and in a variety of ways.

The second set of difficulties result from the inability to move away from the expert, outsider knowledge base and to develop a new insider knowledge base. It can be argued that the sociology of chronic illness itself developed through the realisation that the bio-medical model of health does not adequately account

for the significant effect of chronic illness on the lives of the sufferers and their families. This development heralded the emergence of a body of research focusing on the experiences of 'insiders' as a challenge to the views of 'outsiders. The same developments can be seen within the sociology of disability where it is suggested that broad studies on disablement often fail to look at the subjective experiences of the people with the disabling condition (Anderson & Bury, 1998). However, within the disability movement, this development has been taken further in the creation of a social model of disability which disregards the need for any bio-medical or biological input at all. Juvenile Batten's disease is a genetic condition with profound, degenerative symptoms which require increasing medical management over the course of the condition. The uncertain nature of its symptomatology and trajectory mean that it cannot be fully understood if disconnected from its bio-medical origins. Bio-medicine provides the diagnosis, prognosis and any hope for a cure of a 'normal' life. This means that the social model approach to the study of disability is, in this case, only useful when taken alongside the biological facts of the condition. In relation to data collection this means that bio-medical and lay accounts are intrinsically linked.

The final set of issues that arise in the study of juvenile Batten's disease are problems which could effect researchers studying any rare condition with a limited sample population. There are approximately 50-75 people in the country at any one time with juvenile Batten's disease (Goebel et al, 1999), therefore, meaningful quantitative work is not possible. In addition to this all research done in this area, whether medical, biological, genetic, social or

related to areas such as care, education or family support, are all going to approach this same small group of families and professionals. This means that the group as a whole is vastly over-researched and makes the use of control groups and randomisation of the sample impossible, and re-interviewing problematic and ethically questionable. Further to this, in studying the effects of this condition, one is essentially looking at the whole condition within England and not simply at a sample group. All of the participants are interconnected, whether through the Batten Disease Family Association, seeAbility, charities, schools or care homes. The information collected is inter-linked and the different accounts coincide in many places. This makes the picture created through the data both comprehensive and extremely complex. It also means that I, as the researcher, have played a significant role in the lives of the people that are involved in the study, having been involved across an extended period of time, in a number of ways and in various aspects of their lives, and thus illustrating the need to acknowledge the role of the research, particularly in a study of this kind where the part played by the researcher is significant.

A Summary of the Three Approaches Key to this Study

The following tables illustrate the key aspects of the traditional sociological approach to the study of both chronic illness and disability, the approach necessary when looking at juvenile Batten disease, and the solutions provided through the adoption of a critical realist, ethnomethodologically informed ethnographic approach to the study of chronic illness and disability.

Table 5

Traditional Approaches to Chronic Illness	Juvenile Batten Disease	Critical Realist Ethnography
Challenge to Bio-Medicine	Centrality of Bio-Medicine	allows for knowledge of bio-medicine and family
User-Led Research	Expert-Led Research	Eclectic – information from all sources
Carers as Carers	Carers as Sufferers	Carers as central to research process
Biological Body is Incidental	Body is Central	Body Observed
All Stems from Diagnosis	Diagnosis is Secondary	Explores Identity both through speech and action

Table 6

Traditional Approaches to Disability	Juvenile Batten Disease	Critical Realist Ethnography
User-Led Research	Expert-Led Research	Eclectic – information from all sources
Insider Accounts	Outsider Accounts	Insider observation, outsider accounts
Disabling Environments	Impaired Body	Role of Body Observed
Agency	Lack of Agency	Power Relations Explored beyond surface level
Challenges Bio-Medical Dominance	Welcomes Bio-Medical Dominance	allows for knowledge of bio-medicine and family

Methods of Data Collection Employed

This study uses triangulation of data collection – not for interactionist reasons of the validity of information if collected from a variety of natural and artificial settings – but for the simple reason, common to the study of any subject with an extremely limited sample population, that it is the only way to collect a reasonable amount of information relating to the topic area. Bearing in mind the methodological problems highlighted in the previous section, seven complimentary methods of data collection were used in this study in an attempt to build a comprehensive picture of the experiences of families with juvenile Batten's disease.

The central data consists of seventeen in-depth semi-structured interviews with family members. A loose interview schedule was followed and the interviews covered pre diagnosis through to current experiences, future predictions and in some cases the death of the child. The mother was present in all of the interviews, the father in half and an assortment of siblings participated as and when they were present. This allowed us to get a range of views and experiences from a cross-section of family members, whilst at the same time maintaining some continuity by always having the mother present in the interview. At the time of data collection there was no national database and no specific support group for families with juvenile Batten's disease in England and so families were approached through Health Authorities, Special Schools, Care Units, Support Group, Hospices, Genetics Units, Paediatric Neurology

Departments and Paediatric Neurologists. All people approached were asked to pass a letter to families that they knew and families responded and took part on the interviews only if they chose to do so.

Secondary data was collected in the form of five independent care assessments of the needs of the young people. In addition to this, data was collected from carers, teachers, doctors and support groups. Special schools are widely recognised as a key aspect of the experiences of children and young adults with juvenile Batten's disease and, thus, information was collected from a number of teachers. Two semi-structured interviews were carried out with special school teachers with a particular interest in the education of children with this condition and non-participant observations were carried out in three schools. In addition to this, policy documentation on the wider educational needs of young people with juvenile Batten disease was collected along with written correspondence from a further six schools.

Information was also obtained from carers who were directly involved in caring for young adults with the condition. Five unstructured interviews were carried out and five non-participant observations of care in practice were also undertaken. Further to this I was also able to undertake a two and a half year participant observation of the setting up of a care home caring specifically for young adults with juvenile Batten's disease and other conditions with similar symptoms and life expectancies. I took part in meetings, planning sessions and was involved in the writing of policy documents, the aims and objectives and the care philosophy of the unit.

Another source of information on the experiences of people with juvenile Batten's disease is the medical profession. However, the rarity of the condition, again, means that there are very few doctors in the United Kingdom with expertise in the area, or even with contact with families with the condition. Nevertheless it seemed important to attempt to collect data from this group of people. Letters were sent to all of the main Neurological, Paediatric and Genetic Units in England as well as to all of the Specialists on the list of the Royal College of Paediatrics and child Health. From this initial approach, thirty letters with information in them were received, and I was also able to carry out one unstructured interview with a specialist in Birmingham. Thirty structured questionnaires were then sent out, 17 of which were returned, focusing on the doctors experiences of the condition, what they found most interesting and why they had gone in to the area.

The final data collection took place in the form of a two year participant observation of the setting up of the Batten Disease family Association, a support group for the families of people with all forms of Batten disease. I was involved in the initial meeting and all following committee meetings for the group for a period of two and a half years from November 1998, from which I have collected minutes, agendas and notes. In addition to this I collected newsletters, a wealth of correspondence with families and professionals, the constitution and a collection of politically motivated letters from other support groups and organisation. I also sent out twenty postal questionnaires to

members of the group asking about their requirements and how they would like to see the group develop.

A multi-methodological, triangulated approach, as illustrated above, has allowed the collection of a considerable amount of information from a large number of different sources despite the limitations of studying a very small potential sample population.

Data Analysis Techniques

Qualitative data analysis focuses on uniting theory and method and uses the minimum of quantitative, statistical techniques of data analysis. This means that data is commonly analysed during data collection as well as after all of the information has been collected. Sarantakos suggests that the nature of analysis of qualitative data results directly from the processes employed during the collection of the data:

> "it's process brings together collection and analysis of data in such a way that identifying data leads automatically to their analysis, which in turn directs the researcher to the area in which new data should be sought and identified, in order to be analysed again" (Sarantakos, 1998: 314; from Carspecken and Apple 1992).

The process of data analyses which takes place as a continuous process throughout the data collection phase of the project has been described as the

'Cyclical Process of Analysis' (Sarantakos 1998). This process Involves three stages:

1. Data Reduction – whereby the key themes are identified, and pulled out of the data.
2. Data Organisation – This is followed by the organisation of the data around the themes identified in the previous step. The data is then presented in this thematic format.
3. Interpretation – The final step of the process involves the identification of patterns and trends within the data.

The identification of patterns and trends then guides the researcher to the collection of further data and so the process begins again. This cyclical process is repeated over and over until sufficient evidence is collected to gain an understanding of the phenomenon being studied. This process is also referred to by Miles and Huberman (1994) as 'Pattern Coding'.

This project makes use of the cyclical process of analysis as the tool for analysing the data collected through the fieldwork. This process is located, however in a wider framework incorporating theoretical perspectives on the area being studied and the existing literature within the area. This was done using the technique of thematic framework analysis. This technique allows for the identification of key themes from both the theory and the literature before the data collection is undertaken. These are combined with the themes emerging from the data collection and are used as a framework for

understanding emerging patterns within the data. These themes are then elaborated on as other topics emerge, both through the analysis and further data collection. This allows the themes to be related to all levels of the social world: at the level of the empirical (observed data collection); the processes through which these observed events and experiences are created; and at the level of the real mechanisms which influence the processes and events. Thus the analysis technique being utilised ties in with the theoretical framework being used within this research.

Summary

This chapter started by outlining the reasoning behind adopting a critical realist ontology within this study, allowing us to look at the experiences of families with juvenile Batten disease at the level of the empirical, but also to look beyond that at the ways in which the experiences are influenced by, and influence our current understanding through the literature and theory surrounding this area. A variety of approaches have been used when attempting to study and understand the experiences of people with chronic illnesses and disabilities. We then explored some of the most common of these techniques and outlined the reasoning behind the use of an ethnographic approach within this study. The next part of the chapter outlined the particular methodological problems pertaining to the study of this particular condition but also to the wider study of rare chronic illnesses of this types, and reviewed the techniques of data collection used to combat these problems. Finally we outlined the data analysis techniques used to make sense of the data collected,

and its place within our current understanding of the area of chronic illness and disability within sociology. The reasoning behind the theoretical basis and methods of data collection and analysis outlined in this chapter will become more apparent in the following three chapters where we look first at the experiences of families with juvenile Batten disease at the level of the empirical, and then move beyond that to establish how their experiences fit in with, and challenge, the theoretical framework on which our existing understanding has been built.

5 The Experiences of Families with Juvenile Batten Disease

This chapter presents a picture of the day-to-day experiences of families with juvenile Batten disease. It focuses on the second of the research questions, exploring the experiences of families through a variety of sources. Empirical data is used to build a picture of the day to day manifestations of living with the disease as well as to look at some of the broader issues faced by the families and others involved in the care of children and young adults with juvenile Batten disease. The primary source of the information is the interview data from the parents and other family members collected through the course of the study. In addition, information collected from carers, teachers and through documentation is incorporated, along with the data from the 17 structured interviews with doctors.

It is important to reiterate here that, for the methodological, practical and ethical reasons explored in some detail in the previous chapter, we are looking at the experiences of families with juvenile Batten disease and not directly at the experiences of the individuals themselves. This being said, in the following chapter we will suggest that, in contrast to many of the other more common, less extreme chronic illnesses, simply looking at these experiences in the light of the literature on caring and carers is not sufficient. The experiences that

less extreme chronic illnesses, simply looking at these experiences in the light of the literature on caring and carers is not sufficient. The experiences that these families go through are so extreme that it could be suggested that they too are experiencing the disease, albeit indirectly.

There is a widespread belief that we should build up a picture of the views, expectations and understanding of an individual in order to understand their particular needs and experiences. This idea has been most eloquently argued in debates around the provision of care. As far back as 1959 the Danish philosopher Soren Kierkegaard suggested that care could only be provided through understanding the 'position' of the person you are caring for:

> If you really want to help somebody, first you must find him where he is. This is the secret of caring. If you cannot do that, it is only an illusion if you think you can help another human being. Helping somebody implies you understand more than he does, but first of all you must understand what he understands. (Kierkegaard, 1989, in Ford et al 1996)

In the case of juvenile Batten disease the family is the foremost way of 'finding' the young person with the disease, and in 'finding' them you are also, to some extent, 'finding' the family. This suggests that the two are bound together within the experiences of the disease.

This chapter starts with an overview of sample of families from which the majority of the data has been collected. This is followed by an outline of the main themes to emerge from the data. Each theme is then taken in turn and explored in more detail, using quotes from the relevant data sources to illustrate the findings of the study. In each section the information from the family is presented first and additional information from other data sources is added where relevant. In this way, similarities in experience, as well as possible discrepancies, can be highlighted.

Family Sample Profile

The families that took part in the study were contacted indirectly through doctors, health authorities, schools, genetic units and care homes. Methodological problems and the lack of a national database of people with the disease meant that the sample would necessarily be contingent on ease of contact rather than random. In addition to this, as there is no information on the heterogeneity of experience of the families with juvenile Batten disease, the sample must be taken to be illustrative of some of the issues commonly experienced, rather than representative of the whole. In all, 17 families were interviewed. Fourteen of the interviews were conducted primarily with the mothers and the three remaining interviews jointly involved both parents. In addition to this, in four of the interviews the fathers were present for at least part of the time, and in three further interviews comments were made that were attributed directly to the fathers although they were not themselves present.

Two of the interviews also incorporated the views of siblings who were present for at least part of the interview process.

All but one of the young people with juvenile Batten disease in this sample were part of a family with more than one child. In addition, two of the families had two affected children (one of whom has died – aged 24). Eleven of the young people with juvenile Batten disease in the sample are male and seven are female. Ages ranged from 8 to 27 years at time of interview and the severity of symptoms ranged from exhibiting only the initial visual impairment to being reliant on 24-hour nursing care, unable to walk, move independently or communicate verbally. At least three of the young people have died since the interviews were undertaken. The age at onset of initial symptoms ranged from 3 years (in a case of early onset juvenile Batten disease) to 8 years with a mean of 5¾ years. The age, at onset, of epilepsy ranged from 8 to 18 years and the age at diagnosis ranged from 5 to 15 years with a mean of 10 years. The time between onset of symptoms and diagnosis ranged from the pre-symptomatic diagnosis of a sibling to 10 years.

The sample can also be categorised by the stage of the disease process into which they can be classified. Due to the heterogeneity of the disease this is not a straightforward chronological classification. For the purposes of this study I am using the communication and mobility measures developed in a previous study carried out in 1998 (Scambler 1998). This measure incorporates levels of mobility and verbal communication rather that using more complex, medical measures (Shroijen 1994, Adams et al 1997). The speech and ambulatory skills

are classified using five point scales developed for this purpose and are measure through observation carried out by the family, interviewer, or through documentation from a range of professionals. The scale for speech runs from standard verbal communication at level one, to no recognisable verbal communication at level five:

Level One:	'Normal' Verbal Communication Skills
Level Two:	Hesitant Speech
Level Three:	Rapid, Stuttering Speech
Level Four:	Occasional Recognisable Words
Level Five:	No Recognisable Verbal Communication Skills

Using these standards, 2 of the young people in the study have level one speech, 4 are at level two, 5 are at level three, 4 are at level four and 2 are at level five. The scale for ambulatory skills works in the same way with levels one to five corresponding to a set of deteriorating skills:

Level One:	Steady, Independent Ambulatory Skills
Level Two:	Unsteady, Independent Ambulatory Skills
Level Three:	Ambulant Only With Support
Level Four:	Sometimes Ambulant wit Support, Uses Wheelchair
Level Five:	Full-time Wheelchair User.

Using these standards, 4 of the young people are at level one, 4 are at level two, 2 are at level three, 2 are at level four and 4 are at level five. When the

measures are combined, the sample contains at least two people at each stage of the disease. This gives an overall picture of the sample of families that we are looking at in terms of the makeup of their family, age range and in their stage of the disease trajectory. The majority of the families were living in the southern third of England, due, at least in part, to being clustered around two of the main special schools for children with complex needs of this kind, that can be found in Kent and Devon.

Overview of the Key Themes

A number of areas were covered within the interviews and through the other sources of data collection. These include the importance, and difficulty, of acquiring understandable, comprehensive information on the condition, the experiences of families at symptom onset and also, crucially, at the time of diagnosis. In addition I collected information on the impact of the diagnosis and the disease itself on the family and, indirectly, on the young person. Provision of care was an issue raised and emphasised time and again and is looked at in more detail both in this chapter and, within the realms of the wider literature on caring, in the next chapter.

The information explored in this section is organised around the ten key themes identified within the introduction and addressed again in chapter three. The experiences and opinions of the families will be added to those of the biomedical profession in this chapter and compared with those to be found within the chronic illness literature in chapter six, and the disability literature in

chapter seven. All of the issues mentioned above fall into one or more of the themes. There is some overlap between the themes across the experiences of the families, and issues will be carried through the themes and may be addressed in one or more sections. The themes are as follows:

- Experiences of diagnosis
- The genetic nature of juvenile Batten disease
- The role of bio-medicine
- Implications of a rare condition
- Degeneration
- Living with profound, multiple disabilities
- Living with cognitive degeneration
- Maintaining a sense of self
- Specialist care needs
- The role of the family

The views and experiences of the families will be addressed first and then these views will be augmented by those of the various other people involved in the care of young people with juvenile Batten disease and their families.

One sentiment is echoed across all of the themes and throughout the views of the families. This is the central importance of quality of life and of maintaining a happy, satisfying, stimulating experience for the young person throughout the course of their life, regardless of the stage of the disease which they are in. All

of the parents interviewed incorporated this view as a prerequisite of a successful life experience:

> "We want to see him enjoying himself and basically getting as much out of life as he can." (Mother O).

> "I just want them to be happy, it's very simple really." (Mother D).

> "Our aim is for her to be happy." (Mother E).

> "This is something that we've always maintained, that he gets a good quality of life." (Mother K).

This provides us with a starting point from which we can build a picture of the day-to-day experiences of families and young people with juvenile Batten disease, building on the extension of quality of life and not just quantity of life. The themes are arranged chronologically to the extent that they start with experiences of diagnosis and the specifics of the disease process. The fact that the family is the final theme to be explored is due, however, not to the perceived lack of importance of the role of the family, but due, rather, to the overarching nature of their role, which incorporates, to a greater or lesser extent, each of the preceding themes.

Experiences of Diagnosis

Parental Views

The diagnosis of a child with juvenile Batten disease was universally accepted as a negative event by the families interviewed in this study, not just in terms of the immediate and long-term implications of the diagnosis, but also because of the genetic nature of the condition and the inadequate, or negative way in which many of the diagnoses were handled by the medical profession. The quality of the medical information received at diagnosis was identified as central to the development of the family's understanding, at least at the bio-medical level, of the disease process and its implications for the family. Simply arriving at the stage of receiving a correct diagnosis was not, however, straightforward. Symptom onset was the point at which the families interviewed began to realise that there was something different about them. Differences are immediately apparent with the most common initial symptom being sight loss. This immediately places the child at odds with the majority population functioning in a sighted world. Even before a diagnosis is received parents have to deal with the possibility, and in many cases reality, that their child will need special help at school and possibly even to move to a school for visually impaired children. Parents had to deal not just with the trauma of watching their children lose their sight but also with the struggle to make people take notice. As one mother (K) elaborates:

> I contacted the school and said that I felt she had a problem and she needed some help and asked for her to be on their...er...special needs audit I think they call it. And they said there was no problem

> with her, she was fine and I was being pushy. And then I contacted my GP who said really if it was an educational problem really the school ought to be dealing with it, and he couldn't find anything wrong with her, he did check. So I went back to the school and they said alright and promised to put her on their register and did absolutely nothing. So I went back to my GP and asked him to humour me and send her for a paediatric assessment. (Mother K).

All of this took place before the child was even seen by a specialist. As more symptoms become apparent, the parents have to watch their children struggle to cope and maintain ground with no rationale to link their observed difficulties. Then they have to watch them go through a series of tests. This is a common problem, and the majority of the parents interviewed felt that their children had been subjected to too many tests. In two cases the parents actually stopped the tests because they felt that they were too distressing for the child.

At this point the medical world seems to be of little help to the families. Fifteen of the seventeen families that I interviewed had seen a minimum of four medical professionals before finally reaching the point where they were given the correct diagnosis. The two remaining families were less specific about the process that led to the diagnosis, and so may also have seen a number of people before receiving the diagnosis. Most commonly, families were referred to GPs, Opticians, Ophthalmologists and Paediatricians. In one, not unusual, case, the child was referred to three different specialists for three separate symptoms without links being made. Another mother (G) explains:

> Until he was 15 we were seeing eye specialists and ones for the fits and he had a heart murmur and he was under an orthodontist and I asked the doctors whether his murmur or eyes and his fits were connected and I was shot down in flames. And I was made to feel...I was stupid to even think it was the impression I got. I was just unfortunate that I had a son who was blind and also epileptic. (Mother G).

This was found to be the common experience among the families interviewed, where the medical professionals encountered were frequently found to be unsympathetic and unwilling to listen to the fears of the family. The diverse combination of symptoms and the rarity of the disease do make it a difficult condition to diagnose, however, although a number of tests are available once the suspicion has been raised. Early diagnosis was attributed to the 'luck' of seeing a medical professional who had encountered juvenile Batten disease before, rather than through efficient and effective bio-medical deduction or through professionals listening to parents descriptions of the wider picture. The rarity of the disease means that few doctors have come in to contact with it, although, as one doctor explained, where the disease had previously been encountered it was frequently considered in cases offering combinations of symptoms:

> It is a disease that we consider frequently in the differential diagnosis of severe seizure disorders, intellectual impairment and

> visual impairment. In the last year a number of children have had rectal biopsies or detailed neurophysiology to rule out Batten's disease.

The point of diagnosis was deemed significant, even by the four families interviewed who had suspicions of juvenile Batten disease before they received the official diagnosis. In one such case, the mother (G) had discovered the diagnosis but was refused a test to prove it.

> It was really frustrating...I mean I knew [he] had Batten's ...I wanted to have a name so that I could say [he's] got this. I needed to have a name...nobody was interested, nobody would talk to me. (Mother G).

In this particular case it took the mother eight years from initial symptom onset to get someone to test her son and confirm the diagnosis. In many cases, juvenile Batten disease was not the first diagnosis given. Eleven of the families received at least one mis-diagnosis before they were given the correct information, and three families had two or more mis-diagnoses before they were finally given the correct one. The onset of symptoms sees the families moving into a series of interactions with the medical world, but the diagnosis is the point at which this becomes a formal relationship and the child ceases to be a child with a variety of symptoms and becomes a case of juvenile Batten disease. This is also the point at which the parents are given the news that will

change the course of the future that they had pictured for themselves and their child.

For most of the families, therefore, the medical profession was their first official contact with the disease and their first source of information on the bio-medical aspect of the disease and it's implications for their child. The majority of the families were not satisfied with the quality and quantity of information made available to them, or with the way in which the information was imparted. The most common complaint was that of a lack of sensitivity, as one mother (P) illustrates:

> The neurologist told us, he asked us to make an appointment to come in and told us it. It wasn't done very well, it could have been handled more sensitively. (Mother P).

There was considerable disparity in the way in which diagnosis was handled by different individuals and different organisations, however. As one mother (F) said:

> ...he said "we've had the test results back, [he] is very ill. He is going to end up in a wheelchair and then he'll die." That is all he said to me and it was dreadful. (Mother F).

For another mother (N) the experience was very different:

> The support was excellent. I can't imagine how it could be better really, it was really good. (Mother N).

Most experiences, however, lie somewhere between these two extremes.

The amount of information volunteered was also assessed and only 3 of the families were satisfied with the quantity of information that they received at the point of diagnosis. Almost half, 7 families, were either dissatisfied with the information they received, or received no information at all. Even where information was made available, it was often not in a family friendly, user-friendly, format. Another mother (D) elaborated:

> We had just had the devastating diagnosis and a whole load of information and no one ever talked it through with us. The information in black and white is much more daunting and negative because you don't get any idea of the humanity of it... (Mother D).

The majority of the families now feel that they have enough information, but that they had to compile it themselves, rather than it being offered to them by the medical professionals that they came into contact with.

The experience of diagnosis was examined from the vantage point of practicalities, how the information given, how much information was offered, and whether or not the implications of the disease were explained. I then went on to look in more detail at the impact of the diagnosis on the family unit and

the wider family. Again, it is important to stress here that I was unable to look at the impact of the diagnosis of the individual with the disease because of the restrictions in the collection of data. The reactions to the diagnosis ranged from complete devastation and shock to a very matter of fact practical need to seek more information and assess the impact of the diagnosis. When asked how she felt on receiving the diagnosis one mother simply replied "devastated". Another mother (C) explained the impact of the diagnosis in more detail:

> A wrecker of families, just, it sort of crushes every hope you've got for your children. (Mother C).

Yet another mother (A) talked about the need to 'just get on with it':

> Well [we were] obviously shocked...God, I can't really remember...Our attitude is rather, you know, these things are sent to try you and you get on with life. There's no point in getting all upset about it... (Mother A).

Two of the families that I talked to had to face the trauma of diagnosis a second time as a second child was affected. One father (C) defined the experience as:

> Pretty grim, you see I could have bet on him not getting it, cause they were so different characters...and having read up all this on Batten's disease I thought he won't, keep our fingers crossed and

> then I read that the odds weren't good but, having read up in all these papers that one in four would be a victim, I read that plenty of times and I thought the other two should have a good chance. (Father C).

This quote illustrates the need for information that is accessible and understandable at the point of diagnosis, but illustrates the fact that, even where the information is available, the outcome is uncertain.

The diagnosis did not just affect the immediate family of the affected children. A number of the families talked about the affect that the diagnosis had on the wider family and on the grandparents. The impact spread further afield where families were particularly close, as one mother (D) explains :

> They both have quite a lot of cousins, they have 6 cousins and I wouldn't mind betting that some of them could do with some help as they get older. My eldest niece is 18, so we go right down to a 2 years old niece. It could be that as they see the children deteriorating they could do with some help. I mean we are quite a close-knit family really. (Mother D).

This suggests that support should be made available for members of the extended family as well as the immediate family, particularly where, such as with juvenile Batten disease, the disease is genetic, and there is a possibility that members of the extended family are unaffected but carry the disease.

The final issues looked at in some detail under the theme of diagnosis was the decision about whether or not to tell the child the content of the diagnosis and its implications. The opinion on this was unanimous amongst the families I interviewed. None of the parents that I talked to had told the children what they had got and what the implications of it were. It was a decision that many of the parents made against the explicit advice given to them by the medical profession. One mother (B) explains:

> [we were told] one thing you must never do to him is lie. Imagine telling a 12 year old nipper he might not have that long to live, you know. But I mean lucky for us, um, whether it was because his sight was going first or not I don't know, and all he said to us when we got home was what did they say, and we just said there's nothing they can do, and he seemed quite happy with that. (Mother B).

One mother (K) said simply:

> I lied and I lie.....the misdiagnosis has helped because I say that they still think that it is that...and they don't really know why it is any worse and why it won't get any better. So I just lie through my teeth really. She is easily fobbed off. (Mother K).

There was a universal feeling that nothing could be worse than the truth, and that to tell the children the details of their condition would have a negative effect

and be damaging to their quality of life. It was felt that, in this particular case, honesty was not the best policy. This being said, almost all of the parents I talked to described a policy of omission rather than outright lies. Questions were avoided or answered in an ambiguous way rather that incorrect information being volunteered.

This was the main point on which the opinions of the parents and the professionals that I talked to diverged. There was a strong feeling amongst the professionals, and particularly those who cared for the children once they were over the age of 18, that honesty was the only acceptable policy, and that, as adults, the young people with the disease had every right to expect honest answers when they asked direct questions. This being said, none of the carers went against the explicit wishes of the parents on this issue.

Views of the Carers

One issue that was raised by all of the carers involved in the care of young adults with juvenile Batten disease was that of the need for negotiation and compromise where parental requests did not fit in with the policy of the home. The problem of meshing parents requests and unit policies was mentioned in one of the units in relation to parents choosing not to inform the young people of their diagnoses. It was suggested that this can put care staff into a very awkward situation when they are asked questions relating to the conditions by the young people in their care. It was suggested that there may be a lack of communication between the parents and staff at the onset of care relating to parental choice and unit policies. Care staff felt that they were being put into

an awkward position and forced to answer questions less than honestly, going against their care ethics.

Views of Doctors

The lack of medical professionals with expertise in juvenile Batten disease due to the rarity of the disease, and the difficulty of making a diagnosis due to the combination of symptoms present were mentioned by a small number of the doctors approached. No mention was made, however, of any problems in communicating with parents or other professionals. It is important to note, however, that the doctors approached were asked about their experiences and not specifically about any problems that they might have encountered.

Views of the Teachers

All of the teachers talked to emphasised the importance of having adequate information and a sound diagnosis when trying to teach and care for children with juvenile Batten disease, It was suggested that knowing the diagnosis was the key to working out a teaching strategy for the child. As one teacher explained:

> The main difficulties related to the fact that a degenerative condition contradicts the premise upon which most of us teach, namely that the child will make progress.

Often teachers within the special school environment will have experience of more that one child with juvenile Batten disease, particularly as the children

send to go to a relatively small number of schools particularly because of their expertise in the area. This can lead to ethical problems where teachers become aware of the likely diagnosis of the child before the medical professionals or the bio-medical tests confirm it. One teacher highlighted this problem, as identified by a number of the teachers talked to:

> I was aware of the early symptoms and sudden sight loss that this girl experienced at 7 years old...As a teacher of VI [Visual Impairment] I felt in a difficult position, advising and counselling parents who were already devastated by the blindness. I was not in a position to voice my own suspicions of juvenile Batten disease. It was several years later after I had left the post that I heard a diagnosis was given.

As this teacher demonstrates, expertise in the area of visual impairment and familiarity with juvenile Batten disease can put teachers into an extremely difficult position. If they suggest a possible diagnosis and are wrong the parents have had worry unnecessarily and if they do not suggest it it may be years before the parents find out what is actually wrong with their child. This suspicion of diagnosis before the tests were carried out is common to a number of the teachers talked to who had seen more than one child with the disease through the course of their work.

The teachers approached also talked about the dilemma that they faced having been specifically asked by parents not to reveal the diagnosis to the child.

Difficulties of relating to the child in a truthful way whilst complying with the wishes of the parents were highlighted as particularly problematic. This is a problem common to all of the people that I talked to who were providing care, in a professional capacity, for young people with juvenile Batten disease, trying to combine the needs of the young person with the needs of their family. One teacher suggested that a set decision needs to be made on giving information to the children, and that any information given needs to be explained carefully as it "will naturally frighten some children to talk about death and dying". It was also suggested that all information given to the children must be discussed and agreed with parents beforehand.

So diagnosis was the first point at which the parents were made fully aware of the nature of the condition that their child was living with. This being said, few parents felt that they were given adequate information and a small minority were satisfied with the way in which they were given the diagnosis. In addition, the professionals that I talked to spoke of the difficulty of diagnosing the condition unless individuals had had prior knowledge of it, due to the combination of non-specific symptoms involved. The best guarantee of a well handled diagnosis and being given adequate information appears to be through the luck of ending up at Great Ormond Street Hospital, which has the most experience of the disease, and consequently has the largest number of staff with direct experience of the disease and what it means to the families involved.

The Genetic Nature of Juvenile Batten Disease

Parental Views

The second theme that emerged from the interviews with the parents was the impact that being diagnosed with a genetic condition had, not just on the parents, but also on the wider family. As was highlighted in the previous section, the genetic nature of juvenile Batten disease means that the diagnosis has implications for the wider family, siblings as well as other blood relations. We looked at whether the implications of the genetic nature of the disease were explained at the point of diagnosis, whether the families were given advice about the availability of carrier testing for siblings and others, at the wider impact of the disease on siblings where one or more child is affected and at how parents decide whether or not to get subsequent children tested when they receive their first diagnosis, and at whether genetic counselling was offered to the families.

For the majority of the families in this study, the formal diagnosis was their first encounter with the fact that their child had a genetic disease. Although the facts of autosomal recessive heredity were in the written information that over half of the parents were given, few had the implications explained in full. Where it was explained, often the parents were confused and in a state of shock and so did not fully take in the information that was given to them. One mother (B), for example, spoke of the confusion of taking on this information with everything else.

Q Did they explain the implications of this to you?

A Well not really no. They just sort of said it was genetic, you know, neither partner was to blame, and all I could think of at that moment was what about [his brother], you know, will he get it, has he got it... (Mother B).

The immediate response to receiving this information was a fear for any other children that had already been born. Longer term issues about whether or not to risk having further children, and whether to get the pre-natally tested, and what action to take if any tests come back positive were not touched on because none of the families that I interviewed had had further children with the same partners after the diagnosis.

Once the diagnosis had been given, the focus turned to whether or not to get any other children tested. Opinion was split as to whether this was a desirable option or not. Three of the families were in the position where they had younger children. One of these families decided to have their child tested and the other two decided to wait and see what happened. The mother (D) who got her son tested explained that, as parents, they could not stand the uncertainty of not knowing. As she says:

...so then of course our dilemma was well do we have [our son] tested. But we came to the conclusion that we had to for our own piece of mind, well we needed to know. So we did and around July, we took our time, we didn't rush into it, but then we did and it was confirmed. (Mother D).

This led to the family being in the situation where they knew that the son would develop the disease before he had any symptoms, and knew that everything that they witnessed in their older child would happen, sooner or later, to the younger child. This being said, when asked whether they regretted their decision, the mother (D) said that they needed to know and could not have taken any other course of action.

Conversely, the second family that I talked to said that they decided that they would rather wait and see. When their oldest child was diagnosed with juvenile Batten disease, they had already had two further children. As the mother (C) explained:

> No, we didn't have [his brother] tested....we never had [his sister], we kept our fingers crossed about her, well we thought really...We didn't think [his brother] would get it because he was so different really. (Mother C).

The father (C) spoke of the inevitability of the decline and their realisation that their second child was also showing signs of the disease:

> But, you know, exactly the same problem with the reading, writing, drawing, just everything started to go haywire, so we knew we had to have some tests. (Father C).

He also spoke of his relief when their third child failed to show the same signs as her older siblings:

> by the time she got to 11 we just drew a sigh of relief, what did you used to say, she won't get it because she's good at maths, she used to say, or better than the boys were at maths, but, erm, we just kept our fingers crossed. (Father C).

The only other family in the sample with a younger child also chose to wait and see and were relieved to discover tat their younger child was not affected by the disease. None of the other families in my sample had younger children when the first child was diagnosed, although in one family the affected child was a twin whose twin sister failed to develop the disease.

The families with two children with the disease also had the added trauma of a fear of discovery. Whilst taking the decision, in common with all of the other families, not to tell their children about the implications of their disease, they also had to live with the fear that the younger child would realise that they were developing the same symptoms as their older sibling and so could expect to degenerate in the same way. One mother (D) spoke of her fear that her son will realise what is happening to him:

> ...it scares me sometimes, that [my son] will realise he is deteriorating the same way as [my daughter], and he may put two and two together and that scares me. He takes the mickey out of her

because she stutters, can't get the words out, and it scares me that one day he will realise he is doing the same. (Mother D).

Despite this, both she and her husband have still decided that they will not tell either child what is happening, and will simply do their best to prevent their younger child from guessing what will happen to him.

Even where siblings were not affected by the disease, however, the nature of the autosomal recessive heredity means that there is a fifty percent chance that they will be carriers of the disease, that they will have the faulty gene but will not, themselves be affected. This would mean that, although very unlikely, in the event that they were to meet a partner with the same faulty gene, their children would also have the chance of developing juvenile Batten disease. This led to the issue of whether or not siblings, and other family members, should be tested to see whether or not they were carriers. Although the situation is improving now, some children were told that they could not be tested until they were in the situation where they were in a partnership and deciding whether or not to have children. Other siblings decided that they were too young and would decide whether to have the tests done at a later date. One family did decide to have both siblings tested however, as the mother (A) explains:

...when the children were, erm, diagnosed as carriers, erm, we had a counselling session there and that was just really pointing out what it really meant genetically.....what it did show me was how wide

spread it is and how far through the family it can be extended. (Mother A).

Although the positive results did not have an immediate effect, as both siblings were not planning on having children immediately, it did give them advanced warning, and they were advised to have their partners tested before they decided to have children. In addition, the diagnoses also made the family aware of the number of people who could potentially carry the faulty gene, were all blood relatives of all infected people tested on a routine basis.

Another issue to emerge through the course of the interviews was linked to the availability, or lack of availability of genetic counselling. Only four of the families that we talked to said that they had been offered formal counselling at any time since their diagnosis. And of these, only one family had actually taken up the offer. One mother (F) said:

There were lots of questions that I would have liked to have asked but we weren't given the opportunity. (Mother F).

More than half of the families that I talked to, however, said that they would not have taken up the offer of counselling even if it had been made. As one mother (N) explained:

> I know what I know. I'm happy to pass on what I know to other people but I just don't feel I would get any benefit from it. (Mother N).

There was also disagreement within families, with one parent alluding to the fact that she and her husband had different coping mechanisms and that while she would have welcomed the opportunity to have formal counselling, he did not see any benefit in it.

Again it seems that information is the key to successfully negotiating the fact of being diagnosed with a genetic condition. Families have to take on board the fact of the genetic nature of the disease and the implications that this has for any other children that they have. The then have to decide whether to have siblings tested. Non-effected siblings in turn have to decide whether or not to be tested as a carrier. And, if they are offered the chance, the family has to decide whether or not they wish to undergo genetic counselling. All of the decisions are based on the information that the family are given and that they collect for themselves, as well as the professional advice that they may, or may not be offered. The fact that the information given on diagnosis is, in many cases, inadequate, again raises question about how families are able to cope with make this further set of serious and potentially traumatic decisions. The fact that the implications of the disease have on the family unit is looked at in more detail in the final section on the role of the family.

The Role of Bio-medicine

Parental Views

As was suggested in the third chapter looking at the bio-medical aspects of juvenile Batten disease and the implications of these for families and young people living with the disease, the bio-medical aspects of the disease cannot be escaped. But, what impact does the role of bio-medicine have on the lives of the families? In the interviews with the families we asked about their treatment at the hands of the medical profession, the different treatments that were available, and the role that the family takes in maintaining treatment regimes.

As was seen at the point of diagnosis, many of the families were not satisfied with the way in which they had been treated by the medical profession. Many had seen a multitude of different doctors and all but two of the families had seen a minimum of four medical professionals before they finally reached the point where they were given a diagnosis. Although the families were having a significant amount of contact with the medical profession, few were completely satisfied with their treatment. One mother (K) talked about the disinterested way in which the doctor she was referred to treated her daughter.

> The professor was only interested all the time he thought he had an interesting case, when he thought she wasn't being co-operative – this sounds very cruel and I am probably being very unfair on him, but this is my perception – he wasn't interested anymore. He didn't get the results out of her, thought we were fussing over nothing, and that was that. (Mother K).

Another mother (L) talked of the ordeal that her daughter had to go through whilst undergoing tests, and the unsympathetic way in which she was treated whilst undergoing procedures that were both invasive and uncomfortable.

> They did a test...to try and find out what parts of the eye were reacting, could see light, so [she] had to be in the dark for about quarter of an hour, totally in the dark. She had a swimming goggles contraption, she was totally covered up and she couldn't see anything.....then she was supposed to have these sensors on her eyelids and then flashing lights to record which parts were flashing. Well they kept us waiting for about half an hour and she was absolutely petrified. She wouldn't have the sensors put on her eyes, the whole day was a total waste of time. The specialist didn't want to see us because [my daughter] had been unhelpful, but frankly, with the benefit of hindsight, I don't blame the child. It was appalling. (Mother L).

The poor treatment that many of the families were faced with when confronting the medical profession made them wary of using the services. However, whilst the majority of the care can take place within the home, there are times when the young person has to be admitted, usually for a series uncontrolled epileptic seizures (status epilepticus) or a surgical procedure such as a gastrostomy. Although there was an acknowledgement that there were groups of doctors in particular institutions who had more knowledge about the disease and were

better able to relate to the parents and answer their specific questions and fears, parents still encountered failures with basic levels of care when they were forced to use bio-medical services.

Most problems were encountered when the children or young adults were admitted to hospital and were faced with a whole staff group who were unaware either of the implications of the disease, or the level of awareness of the young person. When referring to her decision not to tell her son of his diagnosis, one mother (B) explained the difficulty she had in maintaining this policy in the face of hospital staff who did not, themselves, realise the implications of it.

> Ever since the diagnosis, whoever we've met or whoever we've had to see we have always said he doesn't know about the condition. They must never mention it in front of him...now one junior doctor...his usual doctor wasn't there...and he said have you got any other diseases in the family. This was all in front of [my son], and I said stop! And when he was in hospital this time ...one of the nurses said, when he was laid there, and he was sort of half conscious, and she said what is Batten's disease exactly? Now even if [he] didn't realise it was about him...I don't want him to hear that because he might hear it again somewhere and put 2 and 2 together, because he can do. (Mother B).

Other problems were encountered when levels of care were inadequate and specialist needs were simply not recognised or taken into consideration. One mother (C) experienced a distinct lack of knowledge and insight on the part of hospital workers:

> In hospital you find problems, you go and find a cold coffee plonked down beside him, nobody bothers to tell him it's there, and he couldn't drink it out of a normal cup anyway.....In fact one of the nurses said, in front of [us], she said we're moving you, [he] was nearest to the glass at the nurses station, she said we're moving you. This was halfway through the week, he'd not just been admitted. We're going to put you by the window, that will be better because then you'll be able to see everything that's going on. She didn't even know he was blind. (Mother C).

Basic misunderstandings of this kind were not one off events, but were experienced by a number of the families that I talked to. This being said, however, information and treatment is slowly improving as the profile of the disease is raised. The families that I talked to with younger children, children diagnosed in the last four or five years, were slightly less likely to encounter this level of ignorance about the disease as more people have heard or it.

All of the families that I talked to, perhaps because of the general lack of information that they often received from the medical professionals that they encountered, sought information, independently from the medical profession,

on the treatment options available to them and any possible developments in treatments or cures. In the last three years three conferences have been held where parents have invited speakers from both the medical and bio-medical research communities to update them on the latest developments. As bio-medical advancement offers the only cure, parents are unwilling and unable to turn their backs on the profession no matter how inadequate the treatment that they receive might be. One mother (D) explained that any chance had to be taken, however small.

> ...we also went to an RTMDC conference where we saw Professor Bennet who recommended fish oils, so they are on fish oils as well, on the basis that they don't do any harm, and we'd feel pretty sick if, in two years time or whatever, we discovered that they would have done some good and we hadn't put them on them. They don't mind them, they are part of their lives now. (Mother D).

In many cases the families also built up a good relationship, over time, with their local General Practitioners, who gained an increasing amount of knowledge about the disease as time went on.

Although there is no cure for the disease, treatments are available for some of the symptoms. This may involve the child taking a range of different pills and medications daily as well as needing regular physiotherapy and other therapy treatments. In many cases this entails parents taking on a whole range of new

skills as they try to cope with the demands being made on them in terms of providing this care. One mother (B) experienced common problems:

> They were giving me medicine and I was supposed to give him 11 mils of this medicine four times a day, and I've only got a 5 mil spoon. How do I give him 11? You know, so, and I could never give it to him at, say like 10 o'clock, 1 o'clock, 4 o'clock, cause he was never here, so it was just get it when you can. So in the end I used to give him four doses of 10 mils and then when he's go to bed I'd give him 15. (Mother B).

Difficulties with medication was only one of a multitude of problems faced by one mother (G).

> I can only do so much, like physiotherapy, you know, I'm limited with the room...Sometimes he won't eat in the morning so I put him on the pump...I have to take the tube off and syringe it through and clean it, you know...I do everything, all the nursing care...I just picked it up as I went along. It's like when [he] started having fits, we went to the doctor and he was in hospital. I just get on and do it. It's like when he had his gastrostomy done, I was shown when he had it in the hospital, this is what you do...I was just shown once, you know, and I learnt it there. (mother G).

These problems are faced by parents who care, either through choice or necessity, for their children at home. Other issues are faced when the care is taking place away from the family home. These issues are explored in the sections on specialised care needs and the role of the family.

Views of Carers

Carers come into contact with the biomedical aspects of juvenile Batten disease predominantly when nursing care becomes an essential part of the daily management of the disease. The need for nursing care was identified by carers in two of the units visited as essential to the care that they provide. The carers in the other units provide care to the point at which nursing care becomes essential. This puts a time limit on the length of time that the young people can stay in the care units where nursing care is not available as a matter of course and where the units are unable, or choose not to provide nursing care. Aside from the final stages the carers only mentioned the bio-medical aspects of the disease in relation to the services which the clients needed to access such as ophthalmological check-ups, physiotherapy and speech therapy.

Views of Doctors

The role of doctors in the day-to-day care of young people with juvenile Batten disease is minimal. Aside from the initial examinations and diagnosis, contact seems to be confined to check-ups with specialists every six months and periodical reviews of drug regimen and the development of the disease. In the words of one Consultant Paediatric Neurologist:

> I see her every six months and in terms of hospital follow up, this particular patient would have a full range of hospital services available to them including ophthalmological care.

This being said, the importance of regular medical assessment is highlighted by the fact that links have been made between doctors and special schools so that the doctor's assessments are built into the regular care provided by the school.

The doctors contacted suggested that they monitor the situation regularly until such a time as more active medical intervention is needed. This generally takes the form of reviewing problems with medication; intervening if the epilepsy gets out of control or fitting naso-gastric tubes or other feeding devices. It is only in the final stages of the disease that more intensive medical care is needed to maintain life, mediate symptoms or eradicate pain

Views of Teachers

Special schools are embroiled in the bio-medical care needs of the children in their care as well as the education needs to the extent that they provide a central point through which the bio-medical needs can be co-ordinated. Through the documentation I was sent it is clear that the support system that is available to provide care fort he students in a typical special school environment is comprehensive, covering, education, social and medical needs.

> At school the children are taught in mixed ability classes and follow a broad National Curriculum. Children have access to a

> school nurse and doctor if necessary and a school Ophthalmologist if and when required. All children are seen by an Ophthalmologist from Guy's or Great Ormond Street Hospitals...A pert-time Educational Psychologist is available to discuss any problems about the child with staff and parents. A part-time Speech and Language Therapist works with these children. Mobility training is taught to the children at school.

It is clear from this that there is a comprehensive support system available within the school for the students and their families. It is less clear from the parents responses, however, how aware they are of the support that is available to them, although the comments on the care provided within the special school system were generally positive.

Implications of a Rare Condition

Parental Views

As was illustrated in chapter three, juvenile Batten disease is a very rare condition, effecting approximately 50 to 100 people in the United Kingdom at any one time. This, in itself, has a number of implications for families trying to cope with the many demands made on their time and emotions through the course of the condition. The limited amount of information available, limited number of people who have expertise in the area, shortage of specialised care skills and small number of other families with which to share experiences and support, added to the fact that the symptoms of the disease will manifest

themselves in different ways in each individual case, means that it is easy for families to feel isolated and to lack the information and support that they need. Two things emerged when looking in detail at the implications of a rare disease of this type on the coping skills and daily lives of the family. Firstly there is the importance of having contact with other families in the same, or similar, positions, and secondly, there is the constant need for more information, not just on the medical aspects of the disease, but also on the practical, day to day reality of living with the condition and caring for a child with juvenile Batten disease.

Almost all of the parents that I talked to emphasised the importance of having contact with other parents of children with juvenile Batten disease. Whilst the reality of the prevalence of the disease means that there are areas of the country with very few families, it was felt that any contact, whether by phone or in person, was better than none. In the words of one mother (F):

> We feel isolated, there's someone somewhere else who feels isolated, there's a girl in...with a kid who feels isolated and somebody else, you know, we are all over the place, all in the same boat and it's just getting us all together really. (Mother F).

Contact with other families in a similar position was important for alleviating feelings of isolation. It was also seen as a useful way of gaining more information about the disease and its effects from people who had faced, or were currently facing the same kinds of problems and issues. Information and

advice of this kind cannot be gained from anyone other than a person who has been through the same event. It provides practical ideas of coping mechanisms as well as possible glimpses of the future. One mother (L) explains:

> ...I think talking to other parents has been a good source of help. And seeing how their children are developing, for want of a better word, and the problems that they had. That has been a great help, it's been a source of a lot of information. (Mother L).

One of the key aims of the Batten Disease Family Association, which was set up in 1997, was to provide a point of contact for parents, enabling them to contact and support one another. The setting up of the group, and the impact that it has had on the lives of the families, is looked at in more detail in the next chapter.

Gaining access to comprehensive and comprehensible information on the disease is another area which becomes more complicated when it is information on a rare disease that is required. Access to information seems to depend on the geographical location of the family, the school that the young person attends and the competency and communicative ability of the medical professionals encountered. The parents expressed the difficulty of compiling comprehensive information, highlighting the need to get each separate piece of information from different places or people. The majority of parents expressed the need for a centralised information centre. In the words of one mother (L):

> I think it would be nice to have...a central point that you could go to and say my child has got this, help, what do I need to do now...almost have a little tick list for you. (Mother L).

or, as another mother reiterated (C), at the very least, access to someone who could point them in the right direction.

> I know it's difficult because the condition effects different children in different ways at different times [but] we had nothing, no guidelines, no – nothing – literally. (Mother C).

'CLIMB' (formally the 'Research Trust for Metabolic Diseases for Children'), 'Great Ormond Street Hospital for Sick Children', 'The Children's Trust', 'Dorton House School', 'The West of England School for Children With Little or No Sight', "Condover School' and 'Henshaws School' were the main institutional providers of information in Britain. Since 1998, however, the Batten Disease Family Association has been taking over that role and provides the families with a centralised point of contact for both professionals and other families. In addition to these organisations, parents researched the disease in libraries, contacted the 'Batten Disease Support and Research Association' in the United States of America, compiled information from the internet and, where possible, contacted other parents, to put together the information that they needed on the disease. Often the information needed was very basic as a mother (D) explained:.

> I don't know what to expect next...I'm blaming everything on the disease whereas I would love to know how much of it is just being a thirteen year old, and I'm not sure anyone can really give me that answer, but nobody has actually said to me I can't give you that answer. I'm looking for honesty, I'm not looking for anything to be, you can't cover this disease up...I'm stunned by the amount of metabolic diseases there are around and how cruel they are, this is evil, it's down right evil...but there is no point in hiding me from the fact. I just want to know what is going on. I'd love to understand what is actually happening inside her body and I don't. (Mother D).

Access to information, therefore, was seen to be a very important part of the process of coming to terms with and understanding the disease, and at the same time was anything but a straightforward process. Practical information was even more difficult to come by. The overwhelming feeling among the families was that there was not enough practical information available and that the little that is available is not accessible through a central resource. All of the information that parents have on the practical day-to-day care of their children has been put together by the parents themselves, often by trial and error, or with the help and advice of other parents.

Clearly the fact that juvenile Batten disease is a rare condition has a huge impact on the amount of information available to the families as well as to the

ease with which they are able to make and maintain contact with other families in the same position as them. The fact that the age at onset and the impact of the different symptoms varies for each individual child compounds these problems. It was suggested that ideally information needs to be available that is individualised to the specific needs, strengths and weaknesses of each child. This being said, however, the general opinion seems to be that and information vis-à-vis the disease, treatment, care and so forth, however general, is better than no information at all.

Views of Carers

The difficulty of caring for a child or young person with a rare condition was highlighted by all of the carers interviewed. The carers in two of the residential units visited suggested that it is difficult to get adequate information about the disease because there is so little information available on the impact and implications of the disease. The carers also reflected the views expressed by a number of the parents, stating that there was very little information on the practical issues of caring for a young person with juvenile Batten disease aside from the information produced by the carers themselves. They produced information sheets for colleagues about the disease and shared the information that they gained through experience in this way. Three of the care units visited had collated all of the information collected on the disease and written additional information based on their own experiences. This makes up the bulk of the information available in Britain at the present time on the care of people with the disease. It does, however, mean that carers have to rely on, and build

up, their own experiences in order to provide appropriate care and are not easily able to access information from other sources.

Views of Doctors

The first, unanimous, point raised by the doctors in the study was that the rarity of the disease means that it is difficult to acquire first hand knowledge of it. Comments ranged from: "Although I have been working in...for almost 30 years, I have never seen a case of juvenile Batten's disease in this area." To "We have never actually seen a case in the past 25 years in this hospital". It was noted, however, that tests for the disease were used more frequently by some doctors who had had experience of it in order to take juvenile Batten disease out of the equation when symptoms were presented. As one Consultant Paediatric Neurologist explained:

> It is a disease that we consider frequently in the differential diagnosis of severe seizure disorders, intellectual impairment and visual impairment. In the last year a number of children have had rectal biopsies or detailed neurophysiology to rule out Batten's disease.

A number of the doctors approached then went on to note that the diversity of the impact of the symptoms of the disease means that a number of cases are required in order to build up an accurate picture of the disease. It was noted that the disease affects each individual in a different way and that even siblings do not necessarily follow the same pattern of disease progression. Experience,

therefore, was seen as the only real way of building up a picture of the disease, but was difficult to gain unless the doctors were based at one of the key paediatric neurology centres such as Great Ormond Street Hospital for Sick Children in London.

Views of Teachers

The rarity of juvenile Batten disease was only mentioned overtly by one teacher who spoke of the difficulty he faced when he deduced the likely diagnosis of one of his pupils before she was given any tests or had it confirmed.

> I was aware of the early symptoms and sudden sight loss that this girl experienced at 7 years old...As a teacher of VI [visual impairment] I felt in a difficult position, advising and counselling parents who were already devastated by the blindness. I was not in a position to voice my own suspicions of Juvenile Batten's Disease. It was several years later after I had left the post that I heard a diagnosis was given.

This reflects the fact that special school teachers working in the larger special schools are likely to have more knowledge about the disease than many of the medical professionals that the parents will come into contact with on a day-to-day basis. How this expertise could be shared is something which has not yet been explored.

Degeneration

Parental Views

The fifth theme to emerge through the empirical data was concerned with the impact of living with a degenerative condition. Three key issue were raised within this area. Firstly there was the difficulty of coping with a diagnosis which incorporates the certainty of a premature death with the fact that the dying process will be degenerative and take place over a period of anything up to 25 years from symptom onset. This is followed by the realisation of the inevitability of the decline and the fact that the means of communicating with the child will also deteriorate across the course of the disease to the point where verbal communication and changes in facial expression will no longer be possible. All of this is exacerbated by the fact that the degeneration of the child will inevitably be compared with the progression of their peers. In at least one case this has led to feelings of depression in the mother (F):

> It's not just thinking of [my son] and the disease that brings it on, like when all the lads he went to school with sat their o-levels and GCSEs and passed them and I think [he] should have been there doing that. When they all learned to drive, all these things, all the boys that he grew up with are still in the village...and I think [he] should have done that. (Mother F).

All of these aspects of degeneration in juvenile Batten disease emerged through the interviews with the families and were explored in some detail.

One unique aspect of the experience of living with a disease such as juvenile Batten disease is the length of time that a person has between the onset of symptoms and the diagnosis of the condition, and the death of the child. Unlike more common disease like Multiple Sclerosis, where a premature death is likely but when it will occur and how premature it will be is unknown, with juvenile Batten disease there is the certainty that death will occur between 20 and 25 years after symptom onset. Seven of the families that I interviewed talked about diagnosis as the point at which the grieving process starts. In the words of one mother (A):

> ....you are grieving from the moment you have the diagnosis, it's a very long grievance, and you go through all the anger, sadness, guilt and everything like that. (Mother A).

There is the idea that the parents are grieving the loss of the child over an extended period of time, two decades, during which they have to both get on with living and ensuring that their child leads the most fulfilling, quality, life possible, but also have to come to terms with the inevitable loss of their child. Another mother (F) took this a step further:

> I actually went to speak to a vicar. I don't go to church, I'm not religious, But I thought, if I went there it would make everything alright. That was in the first two weeks. Now he said life is strange, there will be people who lose children before you, and he said you are so sad you are mourning him now. And you are. You

> start to grieve from the day they tell you. You do. And also it is so dreadful because it is so drawn out and so long a period of time that you have to cope with. (Mother F).

The length of time over which the grieving occurs does not, however, lessen the impact of the actually death of the young person, nor the devastation felt by the family when the inevitable actually happens

The inevitability of the decline was also highlighted. Whilst there is uncertainty about when each of the symptoms will develop, how severe they will be when they do develop and the path that the degeneration will take, there is a certainty around the fact that degeneration and premature death are the ultimate finishing point for children with juvenile Batten disease. The combination of inevitability and uncertainty made it more difficult to predict behaviours on an individual basis, whilst the general patterns could be distinguished. One couple (C), who had two children diagnosed with juvenile Batten disease, explained that there was the recognition of similarities between the experiences of their two children:

> It gets stuck like a record, and you know that nothing else is going to go in, however hard you try, and it's just downhill. (Mother C).

Whilst at the same time there were differences in the individual experiences which made comprehension more difficult:

Q So the progress of the disease was different then?

A yes, absolutely different for the two.....you do know what to expect but it's not happening, you see, that's the thing. (Mother C).

This made coping with the condition even more difficult both on a practical and an emotional level.

One mother (A) talked about the acceptance of the degeneration as a way of coping with the disease and the fact that it has to be lived with over an extended period of time. She talked about the importance of being armed with the facts about the disease and the ability to cope once you know, broadly speaking, what is going to happen:

> .....It's erm obviously a shock but once we'd got all the information about it we felt, you know, we felt more at ease because we knew what was going to happen, and so we just, sort of, got on with life and every deterioration as it came we just took on board and managed. (Mother A).

This highlights the fact that families deal with the disease in different ways and that having the basic facts about the pattern that the disease will take can either lead to an extended period of mourning or can allow parents to develop coping strategies that will enable them to live through the experience.

The last point raised in this section concerns the loss of verbal communication skills. For many families this is the point at which the reality of the seriousness of the condition is brought home to them. They can no longer hold a two way, verbal, conversation with their children and have to begin to interpret what it is that their children want to say to them and how they are feeling and what they want or need. Again this process occurs over a period of time, as in this mother's (P) experience:

> It's getting very difficult with communication, one of the problems is with the deterioration. We have certainly noticed that over this year. This year he has gone from stuttering and not being able to get a full sentence out to erm, starting and then giving up, doesn't even try. Occasionally he just repeats the same 'I want, I want, I want, I want'... (Mother P).

The degeneration of communication skills happens over an extended period of time, from the early teens onwards. This allows parents to develop the skills of interpretation and arms them for the point at which this will be their only way of obtaining an idea of what it is that their children are feeling or wanting. Despite the opportunity for preparation, this loss of communication was recognised, by over half of my respondents, as one of the most traumatic aspects of the disease. Particularly in the final stages where the degeneration reaches the point at which it effects facial expression and the ability to smile.

The degenerative nature of juvenile Batten disease is one of the most traumatic aspects of the disease. From the point of diagnosis, whether this is pre or post symptom onset, there is the knowledge of the eventual outcome of the disease. The inevitability of this outcome, along with the many stages of degeneration that the family will have to experience and cope with along the way, make it particularly difficult. Whilst the inevitability of degeneration may be held in common with a range or other, rare and common, chronic illnesses, the time period across which the degeneration occurs and the severity of the losses make it an unusual case. This aspect of the disease is covered by the carers, teachers and doctors on the following sections.

Living with Profound Multiple Disabilities

Parental Views

The difficulties faced by families attempting to live with, and adapt to the needs of a child or young person with profound multiple disabilities was the sixth theme that emerged through the interviews. This was identified in terms of the particular care needs of this group of young people and the difficulties faced when trying to ensure that the activities and social opportunities available to healthy siblings were also accessible for children with increasing physical and cognitive disabilities. Accessing activities on evenings and weekends that were not linked to special schools was seen to be a particular problem for parents whose children still lived at home. In the words of one mother (K):

> If a child's all right there's swimming clubs, brownies, girl guides, there's all sorts of things that are run of an evening. I don't know of anything in this area where you could have a regular club for a child with a disability, whether it's brownies or whatever, getting that feeling of belonging to a club. (Mother K).

It was even more of a problem where, as in the case of this family, there was a younger sibling without the disease who was able to access all of the community recreational facilities and groups without any difficulties. A number of families spoke about the inclusive, welcoming attitude of local groups who developed around the needs of the child. Two families in particular talked about local Brownie packs that had specifically adapted activities so that siblings could attend together despite one having multiple disabilities.

There was less of a problem with finding activities where the children were boarding at special schools, either weekly or termly. Here the activities were set up specifically for children with profound multiple disabilities. This allowed them to take part in a range of physical and social activities that would not necessarily be available to them in the wider community. One mother (P) explained:

> Physically he's very able...He's beginning to be unsteady on his feet but he does Tai Kwon Do at college and they've said it is

> beginning to help with his balance a bit more, so he's a little bit more stable. (Mother P).

In this particular case, not only was the activity specially adapted for the child, but it was also beneficial on more than a social level, helping him to develop the strength and balance in his legs and giving him more stability.

Activities can fail miserably, however, even where activities are supposed to be set up to cater specifically for young people with profound multiple disabilities. The success of the care and activity provided is dependent on the awareness of the person running the activity of the particular needs of people with juvenile Batten disease and the individual personality and needs of the young person that they are working with. As this mother's (B) experiences illustrate, this was even the case where the activity was in the form of physical therapy provided by the local council.

> When I first started taking [him] up there he had this big sore on his foot and obviously couldn't stand or walk properly. All she would do is lay him on this wedge and as soon as he was on the wedge he was out for the count, gone. And then she started throwing a ball at him, you know, and I was thinking...he can't see it... I mean it wasn't as though there was a bell inside so you could hear it. And she would roll this ball to him and I would think 'he's asleep you stupid woman!' (Mother B).

In this case the professional providing the therapy, although trained to provide physical therapy, was clearly not aware either of the basics of providing services for people with visual impairments, or of the individual with whom she was working.

Caring for a child with profound multiple disabilities within the home caring is complex, requiring adaptations, both of the physical environment, and of the attitudes and lifestyles of the family members. This is looked at in more detail within the final theme, the role of the family. This theme shows that the adaptations and access problems faced outside of the home are even more severe and that, even where activities and facilities are supposed to be adapted and accessible, this is not always the case where the needs of the individual are complex.

Views of Teachers

The impact of caring for a child with profound multiple disabilities, including cognitive degeneration, were referred to by a number of the teachers contacted through the course of the study. Often teachers were required to cope with disruptive and violent outburst within the classroom environment. This is not uncommon and has two outcomes. Firstly there is the need to cope with the outburst itself. One teacher explained that different techniques were used to cope with different children. For example:

> In the case of this particular child, I found it most effective to minimise verbal outbursts by consistently readdressing the content of the lesson rather than the behaviour.

The second outcome of the behaviour was the need to explain the behaviour to the other members of the class:

> In a class setting other children would have to be told that the poor behaviour was not intentional.

This was done so that they might understand that the behaviour was not deliberate and that the child was not aware of the way in which they were behaving or the disruption that they were causing to the class as a whole. In this way the incidents were dealt with and the child was able to remain in the classroom.

Living with Cognitive Degeneration

Parental Views

The impact of cognitive degeneration was the seventh theme to emerge through the interviews. This is linked to the following section on the maintenance of the self, in relation to the young person with the disease. The predominant aspect of cognitive degeneration to be explored was that of memory loss. This was primarily short term memory loss, affecting the ability to recall things that have occurred in the recent past, days, weeks and months, as opposed to events

that occurred years ago. Many of the parents talked about the ability to recall, in some detail, events that occurred a number of years ago, whilst remembering activities that occurred within the school day is a real problem. One mother (C) of a 26 year old man explained the problems of living with someone with only a long-term memory in terms of living with a permanent child:

> ...the repetition...it's still kids stuff that we're hearing about and thinking about, and that is deadly. You can do it for a few years if it's developing, but if it's stuck it's like a broken record..... (Mother C).

She highlighted the fact that in their interaction with her son, the family were forced into reliving and recalling events that happened 15 or 20 years ago over and over again as this was the only way to communicate with him about things that he himself was able to recall. She likened the experience to permanently being the mother of a 7 year old. For this particular family there was a degree of frustration not yet evident in the experiences of the other families that I talked to, both because their son was, at 26, one of the oldest people with juvenile Batten disease still living, and also because he was their second son to go through the disease process and so they had been there and experienced all of this before.

A number of families talked about cognitive degeneration in terms of the frustration of the children in their inability to comprehend what it is that is

wrong with them. They know that something is not as it should be but are unable to work out what exactly is wrong. One mother (L) explained:

> She gets very frustrated. She knows something is wrong but she can't put her finger on it. (Mother L).

Again this raises the question of whether children should be told what is wrong with them and how it effects them. This would allow them to understand why they feel this frustration. Although over half of the families talked about the feelings of frustration experienced by their children, however, none of the parents raised the possibility of informing the children of their diagnosis as an option or a way of explaining the feelings of frustration that they were experiencing.

The remaining references to cognitive degeneration relate to the importance of stimulation and the loss of the stimulation gained through schooling in the latter years of the disease process. Stimulation was identified, by all of parents to a greater or lesser degree, as a means of maintaining cognitive functioning for as long as possible. One mother (O) emphasised the fact:

> [Stimulation] is the vital thing that keeps him going. I can't emphasise enough that stimulation keeps the brain going as long as possible, and that keeps them going as long as possible. (Mother O).

Continued, and regular use of the brain was seen to maximise the chances of maintaining brain activity. Stimulation, both physical and mental, was also seen as a way of maintaining interaction with the young person and maximising their quality of life. The more they were able to get involved, whether actively or passively, with activities, the more they were seen as getting pleasure from the things that were happening around them. This idea can be found reiterated within the ninth theme where we look at the specialised care needs of this group of young people.

Education was seen as a crucial part of the maintenance and maximising of cognitive skills. In the early years of the disease process (approximately ages 8 to 14) maximising educational input was seen as a way of boosting brain functioning. Gearing educational activity to a level where the child is always achieving, although tasks themselves may actually be getting progressively easier, was also seen as a way of boosting confidence and the pleasure of the child. In the middle years of the disease (approximately ages 14 to 18), however, attention was focused on the difficulties of keeping children in school, which is where they receive maximum stimulation, when other disabilities start to interfere with their ability to remain in a formal school environment. The difficulty of managing behavioural problems in a school environment was illustrated by one mother (O):

> He had to leave [school] specifically because he was becoming more confused, he was starting to hallucinate. They already had another young man with the same problems who had really gone

> down hill in a big way, he became completely psychotic, and they just, they were traumatised by the experience. (Mother O).

At this point, where school is no longer a viable option, the parents attention turned to finding alternative sources of stimulation for their children that would continue to maintain cognitive functioning for as long as possible. This raises the issue of location of care and what the best care environment is for a young person with complex physical and psychological needs and a degenerative illness. One mother (G) described the care available to her son:

> ...He's taken out of the wheelchair...he can't sit in it too long because it's built for support and it makes him uncomfortable. Basically they put him on a beanbag, but there's little for him to do. Apart from that, that's all the stimulation that [he] gets, you know, because [he] is the only one with Batten's and they haven't got the facilities. (Mother G).

As can be seen from this example, access to the appropriate environment is not always possible, and maintaining the level of stimulation available to young people at school is sometimes impossible where facilities and expertise are not available. These issues, along with parents' views on the ideal care provision, are looked at in section nine.

Views of Teachers

Through correspondence and interviews with two special needs teachers we can build up a picture of the type of care offered in special schools for children with juvenile onset Batten disease. The general aims are broadly similar throughout the spectrum of schools contacted. It was suggested that the school needs to provide a safe environment in which the children can maximise the skills that they have and achieve even while their cognitive skills are degenerating. A variety of different techniques are employed with different children. The adaptation of the curriculum to the needs of the child is a clear way in which they system is being based around the individual child rather than trying to fit the child into the system. This is a theme that emerges repeatedly through the comments of the special school teachers approached within the study.

Maintaining a Sense of Self

Parental Views

The eighth theme to emerge through the accounts of the families was that of the maintenance of the personality and 'self' of the young person. This is a difficult section in the sense that we are talking about the maintenance of a sense of self without talking directly to the young people who are maintaining their 'sense of self'. I think, however, that the justification for this comes through the fact that in the latter stages of the disease the family provide us with the only means of establishing whether or not the personality, self identity and 'sense of self' of the child have changed, through their understanding of who the child was and who they are now. This is the only way we are able to

truly measure any changes that may have occurred where we are unable to directly ask the young person themself. Care staff my also have a good understanding of what the young person is like at the current time, but will lack the in-depth knowledge of that person across the whole life course that is available to the family. Thus, in order to establish whether or not the young people are able to maintain a sense of self, and the mechanisms used to perpetuate this, we look to the parents.

The first thing that many of the parents talked about was the enduring nature of the personality of their individual children. A number of the parents, of both younger and older children, talked about the strength of character shown by their children in coping with the disease, and the fact that their children would not allow the disease to get the better of them. This was described in terms of the individuality of the children shaping the way in which the disease affected them. One mother (F) talked about the determination of her son to carry one regardless.

> One morning he had the most dreadful fit, he really was poorly, it was when they first started and his face was grey. He sat on the bathroom floor and was so violently sick, and his face was grey. And still he insisted I am going to play my music, and he got on with it. And in the end he said my head is spinning I'm going to have to lay down. He is so, he won't give in, and I think that's what kept him going. He really is independent, he really is, which is good. (Mother F).

A number of the parents of older children talked about their children in this way, as fighting the disease and refusing to let it get the better of them.

From this point two mechanisms for the perpetuation of a sense of self were identified. These are the shaping of present identity on past experiences and the necessity of continuity of care and carers. Again this links with the previous section where we looked at the role of memory loss in shaping the experiences of the young people, and the importance of long term memory. Reiterating and reliving pleasurable memories and events from the last is a way of maintaining self in that the young person can identify with the things that they previously enjoyed and relive the pleasures that they had. This is particularly important where there is little or no short term memory and so knowledge of pleasures are stuck in the past. This is not to suggest that new pleasures should not be sought, but that past pleasures may provide a hint to what future pleasures may be. The experiences that these young people draw on for their interests are experiences from the past, the same as the majority of people, but they are solely experiences from the distant past, the realms of the long-term memory. One mother (H) outlined her strategy for maximising long-term memories:

> ...a lot of their enjoyment comes from memories from the past, and their long-term memory is excellent...their short-term memory is not. And so we've always made sure that people have known what his life is about, and that is what we are beginning to get them to know at [his current nursing home]. You need to know about

> people's background, you need to know what is involved with their background, their likes and their dislikes. Much more so, I've been thinking, than in other cases, because they've been normal, they've seen, they've had sight, they've had lots of experiences and that's what they draw on for their interests. (Mother H).

Continuity of care and carers is important in this instance for a number of reasons. Firstly, it allows the carers to develop a knowledge of the young person, what they like and what they do not like, and to develop an understanding of their personality, through prolonged interaction with the individual on a day-to-day basis. Another mother (A) elaborated:

> My husband and I both feel that the continuity is the most important thing for them... Really understanding the situation and understanding him, getting to know him and what he likes and what he doesn't like and that sort of thing. (Mother A).

Secondly, continuity of care is important because it allows the carers to develop links with the family and to get to know the people who know the young person best. This allows the carers to gain further information about the young person in their care and also enables the family to develop a close relationship with the people they have entrusted with the care of their loved one. Finally, continuity of carer allows for the carers to become part of the longer term memory of the young person. If the same care team provides care for a number of years, experiences will pass into the long term memory of the young person that can

be shared by those that care for them, facilitating a better understanding between the care giver and receiver.

The most interesting point to emerge from the explanation offered by the parents above, however, was the fact that, when verbal communication skills have gone, unless there is continuity of care, the carers are reliant solely on family members to maintain the sense of self of the young person, through their knowledge of their loved one's past likes and dislikes. The family takes on the role of the 'maintenance of self' almost by proxy.

Views of Carers

The importance of maintaining continuity of staff was also mentioned by the carers that we spoke to. They acknowledged the benefits of keeping staff for an extended period of time, and particularly the fact that it allowed the staff to build up a close relationship both with the young person and with their families. They also spoke, however, of the difficulties faced in trying to keep staff members for an extended period of time, caring being one of the most notorious professions for having a very high staff turn over. High staff turnover is particularly found where carers are having to deal with emotionally sensitive issues such as the terminal illness of a child.

Another key way in which the young people are encouraged to maintain their sense of self identity is through the continuation of activities and hobbies that they enjoy. A whole range of activities are available to the young people being cared for at all of the units visited. These include bowling, sailing, canoeing,

horse riding, trampolining and yoga. At least one of the units had a specially designated activities co-ordinator whose main job was to provide the activities that the young people wanted to take part in. He explained the philosophy behind the activities and suggested that the only limitation is the extent to which the young person wishes to get involved. He stated that the activities are based around the interests and abilities of each individual and that a considerable amount of time needs to be spent getting to know the client and their views. This was deemed particularly important where there was difficulty with verbal communication and the clients were unable to communicate their likes and dislikes clearly. He did not talk about the role of the parents in providing information on the young person's likes and dislikes, preferring to approach the individual themselves where possible.

Specialist Care Needs

Parental Views

The penultimate theme to emerge through the empirical data collection was that of how best to provide for and understand the specialist care needs of children and young adults with juvenile Batten disease. The lack of appropriate facilities and expertise was highlighted, as was the fact that access to facilities depended on the area in which the family lived. The location and cost of care were outlined along with the need to find good quality care and the role of the family in that care, particularly where it is taking place outside of the family home. Specialist care needs could be split into four main areas, schooling, post school care, caring for the child or young adult at home and respite care.

Within all of these areas there are debates around funding, location and finding appropriate facilities and expertise.

Finding appropriate educational facilities is particularly challenging when given the combination of degeneration and profound multiple disability and the fact that the education system is based around progression rather than regression. A large number of the families that I interviewed have had to fight to get their children enrolled in special schools with the expertise and resources to provide them with the appropriate education. One mother (K) illustrated the problem:

> We were given enormous opposition by the LEA and I think that was a disgrace...we wanted her at a special school...but the LEA said their policy was integration and she had to go to a mainstream school which is four times the distance from here. I felt quite aggrieved that they made it so difficult because it was obvious from the start that mainstream was an idiotic suggestion. (Mother K).

At the time of data collection one family (M) was in the midst of a fight with their Local Education Authority to force them to provide financial support for their child to be educated at a special school which was just over the county boundary from them. The mainstream school that she was at was inappropriate.

> It is not set up for people who are visually impaired and she is the only visually impaired child there...She does not feel as if she fits in

there because she cannot join in things because she can't see what is going on. (Mother M).

In this instance the LEA was forced to back down after a considerable fight because all of the medical opinion backed the parents claim that special schooling would be best for their child.

Two of the parents highlighted that importance of recreational activities and opportunities offered to the children as well as educational facilities. They point out that in the early teens many of a child's recreational activities come from the friendships that they develop in school. If a child has difficulty making friends because they are unable to keep up with their class mates then they are losing out on this essential source of companionship and may end up with no social life at all. This is seen as an extremely important aspect of special educational provision by the majority of the families that I talked to. All of the families that had younger children stated that statementing, education and finding the right school for their child was the biggest problem that they were facing at this point in time. Many LEAs are currently trying to move away from special school education to a more 'inclusive' mainstream educational policy for all children, although many parents see the reasoning behind this as suspect. As one mother (K) said:

Medically all of the arguments were for a special school placement but I think it's just all down to funding. (Mother K).

The pressure parents face in making this decision is exacerbated by Local Education Authority dictates and financial constraints beyond their control.

The lack of places offering appropriate residential care for young adults with juvenile Batten disease is the next problem to be faced by the families. By the time the child is ready to leave school there has often been a serious deterioration in their condition and they may have problems with communication, mobility, feeding and continence in addition to the cognitive problems, visual impairment and epilepsy that they will have previously had. At the time of data collection two of the young adults whose families were interviewed were living away from home in residential accommodation and four were living at home. Of these four, two were back at home through parental choice and two were living at home because there was nowhere locally that offered appropriate care. Although the situation has changed for many of the families that I interviewed, it is their experiences and feelings at this point in time that I am discussing because any post-interview changes are beyond the scope of this study.

There are a number of residential facilities that provide care for young adults with juvenile Batten disease in Britain. At the time of data collection, however, there was no home set up specifically to cater to the needs of the group of young people. The only home that had been developed for this purpose closed down in early 1997. The need to find appropriate, long term, residential care for an adult son or daughter is not something which more people will ever need to consider and yet it is central to the lives of those parents who are trying to

care for children with juvenile Batten disease and other conditions which require long term care. Again it is an area where parents often have to fight against Local Health Authorities and Social Services. One mother's (A) story illustrated the problem:

> They obviously are looking for the cheapest option and we always had to fight, but never as much as we had to fight this time, to get him into somewhere we know where he will be looked after properly. (Mother A).

This is another stage in the disease where parents are finding that they have to fight for things which other people would never even have to consider, and should be taken for granted. Even where homes are available and offer the required levels of social and nursing care, the parents were not always happy with the role that they played in the care of their children outside of the family home. One mother (H) talked of problems in her son's residential home:

> I would like to be more comfortable... when I go into the home I don't always feel comfortable because I feel I shouldn't be there... It would be nice if you could go in there any time of the day and be made to feel you're welcome, only too pleased to see you, instead of being made to feel uncomfortable. (Mother H).

This mother was particularly upset with the attitude of the home in which her son was living. She was generally happy with the standard of care that he was

receiving but felt that she was not welcome, seen as interfering and should not visit as regularly as she wanted to. When asked what she felt her role was in the care of her son she replied:

> I don't think I've got one. Except my daily visits and when he comes home. But I feel they don't listen to me... I was so upset the other time I cried for a week... it made me ill. (Mother H)

She was on anti-depressants which she attributed directly to the 'situation' with the care home. Parents roles in the care of their children, both actual and desired, are looked at in more detail in the final section.

Residential placements are not an option for some of the families that I interviewed, either through choice, or lack of appropriate local residential facilities or, in one instance, due to lack of funds and the local authority's failure to take into account the needs of the family where residential care was favoured. Lack of appropriate residential placements may mean that families are forced to consider providing care at home even where they feel it is detrimental to the quality of life of both their child and themselves. One mother (E) spoke of problems finding appropriate care for her daughter:

> We are looking and there's a possibility of her staying home here. We don't know whether that would be the best thing for her really because she's used to having a lot of people about...so there are options but I don't know how good they are. There are places but

> they don't do nursing care. When she starts she won't need nursing care but they have to be able to handle it or she will have to move again when she does need it...There's a children's hospice that she can go to but that's just for respite care. It's not permanent because really it's for children and she knows that too. (Mother E).

Two of the families interviewed were extremely eager to find appropriate residential accommodation for their grown children but felt that this may not be possible. The father quoted above felt that he and his wife may have to provide care at home if they wanted their daughter to stay within a reasonable distance of the family home because they were unaware of any residential options locally. In the two years since the interviews were carried out, however, an appropriate home has been found that is close to the family home and will provide care for their daughter right up to her death. The other parents who described themselves as reluctantly caring for their son at home were unable to get a residential placement because their local authority refused to pay for residential car until such a time as nursing care was necessary and care could no longer be provided at home cost effectively. The parents, in this particular case, felt that their quality of life was poor and that a residential placement would provide them with the respite that they needed, provide their son with the care he required, and allow them to enjoy the time that they had left to spend with their son.

Two of the families had taken the decision to care for their children at home when they were no longer able to continue full-time education. For these

families it was essential that they had access to day centres with appropriate facilities and, where possible, knowledge about the particular needs of young adults with juvenile Batten disease. One of the families (C) was extremely happy with the day centre they used.

> The day centre here is marvellous, there are marvellous staff, they have a lovely, varied programme. They have very [nice] clients, not difficult people to be with, and the staff are absolutely... couldn't wish for better really. And he is very happy. (Father C)

Two other mothers, however, talked of inappropriate facilities or client groups, lack of specialised knowledge and lack of qualified nursing care in the centres that they were forced to use. This put further strain on the families as they had to fill in the deficits left by the day centres, particularly in relation to medication.

The disease trajectory of a child with juvenile Batten disease can be illustrated through the various stages of care requirements that the child goes through. In the initial stages there is the transition from 'normal' to 'special' educational needs and the possible move to a special school. This is often followed by a transition to residential education with the children boarding at the schools during term time. The next major transition period come where the child, or young adult, is no longer able to remain in full-time education and must leave the special school. At this point the families face the decision of whether or not the child should be cared for at home or within a residential care unit. The final

stage of the disease care process comes with the increased need for nursing, and eventually, terminal care. Only one of the families that I interviewed had been through the terminal stage of the disease at the time of interview. They talked about the final stage of the disease in relation to the experiences of their elder son. The remaining 16 families felt unable to talk, or even think, about this point of the disease. It was discussed only in so far as one mother referred to the fact hat her son would remain in his present residential accommodation for the remainder of his life. It seems to be the case that this is something that is not considered if at all possible until absolutely necessary, although three of the parents of older children did refer to the fact that increased nursing care would be needed and that they did not want their children to be moved around if possible at this stage of the disease.

The importance of the specific care needs of children and young adults with juvenile Batten disease cannot be underestimated. Throughout the course of the disease parents have to make decisions about the kind of care that their children are receiving and the location in which that care should be received. Often they have to fight to gain the type and location of care that they feel best caters for the specific needs of their child, and in all of the cases studied, this leads to at least one period of time where the child is living away from the family home. This is particularly difficult for parents that know that they only have a limited amount of time to spend with their children, and that, for that reason, every day that they have is precious.

Views of Carers

The importance of communication was unanimously voted as the key to the successful running of a care unit. This refers to communication and information sharing between the young people, staff, families, medical professionals and anyone else involved in the care of the young person. The carers stressed that the families must be involved in the care and that, where there was residential care involving significant geographical displacement, sharing of information between families and care staff may be the only was of involving families between visits. The care staff interviewed were very aware of the pivotal role that played in the life of the young person in their care and also in the lives of the other family members.

All of the carers interviewed stated that they were happy with the care that they are able to provide in the course of their work. The carers worked in environments ranging from a large school to a small group home built to accommodate six adults. The carers stressed the importance of building up a good knowledge of the young people that they are caring for; maintaining good communication with the parents; the provision of nursing care where needed; and the provision of a range of activities stimulation and exercise. All of the carers spoke of providing each of these aspects of care to the best of their abilities. It was clear from the interviews with the carers that it was widely felt that the care being offered in the units visited was appropriate to the needs of the young people being cared for and their families. This was interesting as a number of the parents who expressed dissatisfaction were referring to the one or more of the care units visited. This suggested a discrepancy between the

views of the carers and those of the family on the appropriateness and quality of the care being provided, and the quality of the communication between the home and the parents. None of the carers expressed any problems with the care that they provide and the problems highlighted by the parents were not mentioned. It is unclear whether this is because the carers were being interviewed within the workplace and so felt unable to criticise the practices being carried out there, or because they feel that the care being provided at the units in which they work is, on the whole, satisfactory.

Views of Doctors

When talking about the specific care needs of young people with juvenile Batten disease the doctors approached focused on the importance of multi-disciplinary teamwork in providing successful care. One Consultant Child Neurologists highlighted the fact that the school is often the first place to pick up on the visual impairment of the child and that it is through them that the ophthalmologist gets involved, and that often the neurologist is brought in through the ophthalmology assessment. This would suggest that successful diagnosis and treatment depends on multi-disciplinary communication and information sharing.

Views of Teachers

The teachers approached through the course of this research were very aware of the expertise that they held in this area, many of them having come into contact with more than one child with juvenile Batten disease. They stressed the importance of being aware of their pivotal role in the lives of the families for

the period of time where the child is attending their school, and often living away from home for the first time. The importance of maintaining good and open communication with the parents was stressed by all of the teachers, both in face-to-face interviews and through correspondence and the educational plans I gained access to. It was stressed that parents need to be made to feel welcome to visit the school at any time and that staff should be open to discussing the needs of the parents as well as those of the child.

The teachers in the study gave a comprehensive description of the care that is currently available within the special school system. It is clear from this that, if the system is working as it should be, both the needs of the child and their family can be met. However, the special school makes up only one part of the care provision for the child and a comprehensive communication system needs to be in place for the school to work with the funding bodies, medical bodies and other interested parties in providing a full programme of care for each child. Again it's useful to note that the care provided by the special school system will most often end before the child enters the final stages of the disease and so issues of medical care are only touched on at the end of the placement.

The Role of the Family

Parental Views

The tenth and final theme to emerge from the empirical data relates to all of the preceding themes and encompasses all aspects of the daily lives of families with juvenile Batten disease. This theme concerns the role of the family in the

day-to-day lives of children with juvenile Batten disease and the impact of the disease on the family unit, both in terms of individual members and as a whole. As suggested in the introduction to this chapter, it can be suggested that this is the most significant aspect of the empirical data because the families of children with juvenile Batten disease can be viewed as more than carers and even as sufferers of the disease, both directly as carriers, and indirectly, through the impact of living in an 'abnormal' family. This section incorporates the views of the family respondents on family life and the maintenance and breakdown of 'normal' family roles. We also look at specifically at the impact of the disease on unaffected siblings and on the wider family before looking at the views of the family on care environments, family roles in care both within and without the home, and the role of respite care and self help groups.

As previously stated, juvenile Batten disease, in common with other more common chronic illnesses, affects the wider family and not just the affected individual. All of the families that I talked to spoke of the difficulty faced in trying to keep a 'normal' family life going whilst caring for a child (or children) with juvenile Batten disease. One mother (B) explained:

> We used to say we'll plod on the same way as we always do, you know, and just treat them as a normal child, cause I thought, like, you know, well if you don't treat him the same as you always treated him he's gonna suspect something, and know something's wrong. And then you've got all that to explain, so you know, I mean if I though he was getting a bit loud with his music or if he

was doing something he shouldn't be doing I'd be shouting in the same way I always did. (Mother B).

This was a view reflected by a number of parents who talked about the difficulty and frustration of trying to carry on as normal. One mother (O) explained that even seemingly straightforward, spontaneous decisions become difficult:

You can't make any decisions, if we were normal people we would just say, oh we'll have a snack instead of having a main meal, we'll have a snack, you can't, it's just routine. Just a steady, boring routine. (Mother O).

These difficulties are compounded when the child is living outside of the family home and has limited verbal communication skills. One mother (F) talked of the struggle to keep up to date with the life of her son and to keep the family communication lines open.

I'm trying very hard to get them to keep a diary for me just so I can have something, he comes home, mainly some weekends at the moment, and I like to know what he has been doing and he can't tell me. (Mother F).

Problems of communication, role maintenance, spontaneity and discipline are further complicated when the needs of the other family members also have to be incorporated into family life, as another mother (I) illustrated:

> However badly you think you're doing, it's not surprising that you're doing badly because it's very hard to keep your end up and say to cope with a normal member of the family who has needs as well...the thing just sort of afflicts your whole existence really. (Mother I).

One mother (D) talked about the need to protect the time she has at work because of the release it provides and the chance to be a 'normal' person away from family roles and responsibilities.

> ...work to me is very important, I need the money and I need the independence. I've got to think forwards and I can't be at home all the time, I'd just be finished as a person quite frankly. I've got to keep my life going as well, so a little bit of selfishness here but I think I would be a sad woman if I tried to keep them at home for too long. (Mother D).

The struggle to balance multiple roles and pressures and to maintain a sense of self results, perhaps unsurprisingly, in acute, and sometimes terminal, strain on marital relationships.

The nature of juvenile Batten disease as a genetic disease that is passed down through the families of the two parents will mean that the families may feel themselves to be deviant. This, along with differences in the coping mechanisms employed by different partners, can lead to marriage break-ups and to problems within relationships. Two of the parents that I talked to ascribed the break up of their marriages directly to the stress of having to cope with the implications of the diagnosis. Another woman admitted to having problems within her relationship due to the different ways that she and her husband have coped with the disease and the implications of it. She explained that her husband had always felt that he would continue through his children and so the shock of finding out that the genes that he was passing on were faulty had had a big effect on his self-identity and the image he had of himself, both in the present and in the future. Half of the women interviewed are no longer with the father of the affected child. This is a figure that is higher than the national average for divorce, although whether this is due to the disease or to an unrepresentative sample is impossible to say. In many cases this means that one parent, usually the mother, is left to cope with full-time care of a child with complex needs alone, as in this mother's (D) case:

> My husband left home in June last year, there again you have got that hopping into the equation as well. I see him on a regular basis but it isn't the same , I do miss him. (Mother D).

As this mother stated, the absence of the father on a daily basis, even where he still plays an active part in the rearing of his children, leaves the mother isolated and vulnerable to the stress of coping with a potentially traumatic situation.

Juvenile Batten disease also takes a toll on the wider family. One mother (F) talked of difficulties in getting her parents and siblings to cope with the genetic nature of the disease:

> They are just ignoring it basically. (Mother F).

This kind of attitude can lead to the affected family feeling even more isolated and stigmatised. Most of the comments in this section, however, related to the effect of the disease on siblings of the affected child (or children). The parents talked of making judgements on how, when and how much to tell younger siblings about the situation, as one father (C) explained:

> I don't think we ever took her aside, she never really knew what her two brothers had got, well she might have done, but we never told her. We didn't want to worry her really. We did think of sending her away to boarding school to get her out of the way, out of the hassle with the boys, it was mostly playing on her concentration. But she doesn't seem to have any regrets about it. (Father C).

While boarding school may seem an extreme response, a number of the families highlighted the stress of trying to ensure that all family members had

adequate attention. The same family focused on feelings of guilt where they were aware that younger sibling had missed out in some way.

> She does always remind me that I didn't teach her to read...but when she was four, that was the time when both boys were at their worst...and that was the time when she was neglected really. (Mother C).

> ...She has never had any fun, we've never had any fun. We should have had lovely seaside family holidays, playing cricket on the beach, but we have never been able to do that. (Father C).

The reflected the perception that the family was, in some way, deficient because so much of the energy and attention was focused on the one child to the detriment of the other family members.

The reactions of siblings to the implications of the disease were also mentioned. One mother (A) spoke of the difficulties her daughter faced in coming to terms with the fact that her brother was affected while she was just a carrier. She talked of feelings of frustration and guilt and the need for counselling.

> The only one who needed any, well it wasn't really support, about Batten's particularly was [his] twin sister. She goes to therapy, she has had various problems and one of the roots of her problems is the fact that [he] has Batten's. She finds it very difficult to deal with

> being a twin...but she was the only one who needed counselling and she decided herself that it was very helpful. She still needs to talk to people about it, it helps her. (Mother A).

This was not the only family where the siblings were having problems coping with the disease. Where younger brothers and sisters were involved the parents spoke of feelings of embarrassment and stigma with younger brothers and sisters resenting their older, affected, siblings because of their difference. One mother (K) elaborated:

> [Her sister] is a problem really because she is struggling, particularly now in the last couple of months, she hates being different. She says I hate having a blind sister and she doesn't mean that in an unkind way, I think that she just means that she hates being the different one...she suddenly stopped having any interest in inviting friends home, she just shied off all the time. She just didn't want to know about it...Eventually she did admit that she hated having friends when her sister was here because here sister spoils all the games and she just feels embarrassed. (Mother K).

Again this illustrates the wider impact of the disease on the family as a whole.

Juvenile Batten disease has an impact on the family of the affected child in both the home environment and in any alternative care environments in which the child may live. The role of the family in the care of the child or young adult

was looked at in some detail. A number of the families talked about their experiences of caring for their children within the family home. A number of the parents had made contact with each other, allowing them to share experiences of care and the acknowledgement of some of the difficulties that they would face. One mother (L) talked of the experiences of another mother:

> Doesn't know how she does it really, never thought she could but she does, she copes, it's her son and she cares for him. Sees it as instinct really. (Mother L).

In this case both parents were caring for their children at home and felt that this was the most appropriate environment in which to provide the care that their children needed. At least one family (C), however, were restricted to caring for their adult child at home, not through choice, but through a lack of funding for an alternative placement.

> Well we can't afford [care home name] really but if anything happened to us I would be worried if there wasn't an alternative ready. ...I mean we start to think how long are we going to keep going, and you have to be pretty able bodied to keep going. (Mother C).

This put added strain on the family when planning for the future, as they had to consider financial arrangements as well as trying to ensure that their child would receive the best care available.

Few of the families providing care for their children within the family home talked about worries for the future in terms of the death or incapacity of the parents, as most were caring for children who were pre-teen or in their early teens. For this group one of the most important considerations was access to respite care. All of the families providing care at home talked of the importance of having access to good quality respite care and the need for both continuity of care and continuity of carers. One mother (P) talked about the importance of trust.

> Parents are happier to hand their loved one over to someone they know. At the same time the young people are happier of they are left with someone they know and trust. (Mother P).

Although respite care was deemed important for all parents there are few respite places available for children in Britain and very few which cater specifically to the needs of teenagers and young adults. Thus parents often had to make do with a small amount of respite and often had to travel considerable distances to access the facilities. One mother (I) talked of the logistics of making use of the respite provision that they were allowed:

> I really don't think that 15 nights a year is enough. I mean we can't even use the hospice really for one nights stay because it's an hour and a half there and an hour and a half back ... and by the time

> we've settled him in and gone through his care plan ... this just isn't practical for one night. (Mother I).

This mother talked about the importance of respite care because it gave the family a break from the constant needs of the affected child and allowed the siblings to have the undivided attention of their parents for a period of time, and the parents to have some time together as a couple. She felt that this was an extremely important part of the maintenance of the family unit, both for the parents as a couple and for the other children. This view was echoed by all of the parents who were caring for their children within the family home.

Different issues arose when the families were questioned about the impact of having their children cared for in an environment outside of the family home. In the latter stages of the disease, one of the most difficult adjustments for the family is the need to maintain a sense of family, and family roles, in an alien environment. Parents talked about their expectations about the care that should be provided and their roles in that care. Although some of the parents were not totally satisfied with the care that their children were receiving, one mother (N) did admit that she might be difficult to satisfy:

> Nowhere is ideal and nowhere can be... you can never expect other people to do things to the same standard as you do. It's just the difference between being a parent and being a member of a medical or a care team. (Mother N).

This demonstrates the need, expressed (if subconsciously) by a number of the parents, to separate the care given by 'others' from that given by parents. The role of the parents was seen in some ways as sacred. In some cases this resulted in parental expectations that were almost impossible to meet. Another mother (H) explained:

> They kept saying to us 'what do you want?' and I said I want a substitute mum. I want another mum to take over when this mum has had enough. (Mother H).

This would be the best possible alternative to providing the care for her child herself.

The difficulty of maintaining the roles of parents in an alien environment was highlighted by all of the parents with children living away from home. In the words of one mother (F):

> I want day to day information from everyone involved with [him]...yes I like to be involved with [named school], social services, anybody coming into contact with him, because he's my child. (Mother F).

Some parents, as with the next mother (A) were happy with the amount of information that they were given and the role that they played in the care of their child:.

> They refer to me if there's any change in his care or I request any change in his care then they will take that on board and it's all written into his care plan ... They wouldn't do anything to him without me knowing about it. (Mother A).

The majority, however, as another mother explained (H), expressed varying degrees of concern or alienation:

> I sometimes feel they don't want the parents around... I don't think they ever actually stop to think how the parents are feeling. You see they are caring for this severely handicapped lad and it's just a job to them ...not an easy job... but I don't think they ever sort of try ad put themselves in our shoes. I think they don't just stop and think how they would feel if it was one of their children. (Mother H).

This mother felt that the attitude of the care staff threatened her role as a mother. There was disagreement about how the children should be treated, particularly when they were legally adults, and about what role the parents could and should have in the day-to-day care of their children. It was on the point of the rights of the young adult over the rights of the parents that most problems emerged.

It seems clear from the responses of the parents to questions about their role in the care of their child with Batten disease and the impact of the disease on the family as a whole that the parents are having to deal with a multitude of issues, often conflicting with each other, at the same time. They have to weigh up the needs of their affected child with those of their unaffected children and, where possible, make time for their own need as well. The difficulties of maintaining a cohesive family unit under the pressure of chronic illness was looked at in more detail in chapter two, and it seems clear that families with juvenile Batten disease are facing far more potential problems than families without chronically ill children and possibly even more than the average family with a child with a more common, less extreme, chronic illness. These problems are compounded by the fact that the intimate details of family life are open to public scrutiny in a way that they would not normally be both because of the number of outside people coming into the home and the fact that, in the majority of cases, part of the family (the affected child) will be moved to an external environment, taking the relationships that surround them with them. As can be seen, both through the quotes given in this section and by those given in the previous nine sections, the role of the family is challenged and threatened by each aspect of juvenile Batten disease, from the onset of symptoms pre-diagnosis all the way through to the choice of care provision in the final stages of the disease process.

All of the non-parental respondents in the study mentioned the importance of maintaining good relationships between the different groups involved in the care of young people with juvenile Batten disease. The need for communication and information sharing were highlighted, and all groups talked

of the mechanisms that they already have in place to ensure that parents are fully involved in the care that is provided by all of the members of the multi-disciplinary team involved in the care. Whether the parents would agree that these practices are being followed is open to debate.

Summary

Fifteen key points emerged through the empirical data. They are as follows:

1. Diagnosis time depends considerably on whether or not you are referred to a consultant who has come across the disease before. Diagnosis time ranges from months to over 8 years. This may be linked to the fact that all of the families in my sample saw a minimum of three medical professionals before they were given the correct diagnosis.
2. Many parents are not happy with the way in which they are given the initial diagnosis, although no doctors highlighted this as a problem area.
3. Very few parents are offered counselling immediately after diagnosis. Few are offered it at any point after this.
4. Most of the parents feel that they were not given enough information and time to discuss the information with medical professionals.
5. There is a complete lack of practical information about how to cope with the disease on a day-to-day basis available to parents or carers. Information required includes specifications and availability of equipment, feeding, statementing and a whole variety of issues faced on a daily basis.

6. The majority of parents have not been given enough help with finances and have had to fight for special education, money for equipment and alterations to houses.
7. Problems and concerns are immediate and age specific. For example, parents of younger children are concerned about education and statementing and parents of older children are concerned about residential accommodation or the lack of it.
8. The majority of parents do not think in detail about future problems until they can no longer be ignored.
9. The disease is different for every single child that has it. It develops at different speeds and in different ways with symptoms varying in intensity from one child to the next. In addition to this, each person retains their individuality with personal tastes, likes and dislikes that need to be taken into account.
10. The majority of parents identify continuity and stability of care as the most important needs of their child.
11. Parents worry about not being able to give siblings the 'normal' happy childhood that they feel they are missing out on.
12. Many siblings find it hard to adjust to the fact that they have a brother or sister with juvenile Batten disease, they feel guilt, anger and embarrassment.
13. There seems to be little emotional support available to parents outside of friends, family and other parents in a similar situation.

14. Most parents prefer to talk and discuss problems and possible solutions with other parents who have gone through the same things rather than with professionals.
15. The majority of the parents in this sample expressed the view that they were lucky compared to parents of children with other diseases, such as late infantile Batten disease, because the disease trajectory is such that their children would be alive for long enough for them to enjoy a considerable amount of time with them before they eventually die.

Conclusion

In this chapter we have outlined the key aspects of juvenile Batten disease and the impact that the disease has, from symptom onset right through to the terminal stages of the disease process, not just on the individual with the disease but on the family as a whole. The views put forward by carers, teachers and doctors involved in the care have also been incorporated to give a broad overview of the disease, what it means to live with it, and the support mechanisms that are and are not available to the families, carers and other people involved in the process. The empirical data suggests that there is a difference in opinion between the families and non-parental carers on the type of support available and how services are provided. Whether this is because the services are not actually available, parents are not made aware of the support services available to them, parents choose not to take up the support for whatever reason or the support is inappropriate, we do not know. However, what is clear is that better communication is needed between the different

groups so that they can provide information and support and share expertise with each other. It is possible that at least part of the role of liasing and improving communication will fall to the Batten Disease Family Association, the support group that has been set up, in the last three years, by families of children with Batten disease. In the next chapter we look in detail at the literature on caring and the role of families in the care of people with chronic illness and disabilities. We also examine the setting up of the Batten Disease Family Association and the impact of self-help groups on experiences of chronic illness as a whole.

6 The Family and Juvenile Batten Disease: Caring, Self-Help and the Emergence of the Batten Disease Family Association

This thesis focuses on the impact of juvenile Batten disease on the daily lives of the families of children with the condition. In previous chapters we have contended that parents of children with juvenile Batten disease are 'more than' carers, arguing that the experience of the disease, on the part of the parents and the young person themselves, is such that traditional carers/sufferer boundaries cannot be applied. This was further developed in chapter five where the experiences of families were explored in some detail, from symptom onset right through to the terminal phase of the disease. It was suggested that the disease had a profound impact on the lives of both the immediate family and the wider family, due to the profound nature of the disease and it's genetic origins.

In this chapter the literature on caring and self-help is looked at in relation to the experiences of the families. The development of community care and the

implementation of inclusionist policies on schooling and residential care for children and young people are examined. There is an extensive literature on informal care, care within the community and the impact of caring on the carers which is also explored, with particular emphasis on the role of parents and families caring for disabled children. The role of self-help groups in providing support for the families is then examined with recourse both to the growing literature on the role of self-help and through the empirical data collected on the development of the Batten Disease Family Association. Finally we assess the impact of caring on the families, the support that the families are receiving from the Batten Disease Family Association, and the premise that the families are, in effect, sufferers of the disease as well as carers of people with the disease.

Community Care

The increasing emphasis placed, over the last 40 years, on 'community care', or care within the community has had a huge impact on both formal and informal care. The concept of 'Community Care' can be seen in terms of a rhetoric, philosophy and policy, all of which are historically contextual. Henderson and Armstrong (1993) identify two themes within the rhetoric around community care: that care should be provided in a community or home setting, and that it should incorporate the concept of 'normalisation' – that care should be able to be incorporated into everyday life. This original understanding of community care incorporated three aspects of care provision:

1. Care services should be provided in small scale, client centred residential homes within the community.
2. Care services should be provided by professional carers and specialists working within the community.
3. Additional services should be provided on a voluntary or quasi-organised basis – particularly in relation to carer support and advocacy services. (Henderson and Armstrong 1993).

Mays (1997) suggests that the concept of community care is one that is very attractive because it carries implications of living in a caring society where strong social networks will allow people to be cared for within the community. He goes on, however, to suggest that in practice community care is considerably more complex, referring to both formal and informal care in a variety of settings. He suggests community care was originally the idea of formal care carried out outside of institutions, domiciliary care carried out within the community and the care provided by day hospitals and hostels. After 1979, however, there was a change of emphasis with the focus moving towards care provided by the community by informal carers. The original intention of community care, as a means of closing down large scale institutions and providing care for people within smaller groups homes within the community has gradually transmuted into the idea of care provided within the community by the community, with an increased focus on the role of family and friends as carers (Glendinning 1992). Before looking at the literature of caring and the role of informal carers, it is useful to outline the basic tenets of community

care, how it has emerged and changed since the 1950's, and what the requirements on informal carers are.

Up to the late 1970's people many with physical disabilities, learning disabilities and mental illnesses were cared for outside of the community in large institutions, usually located in rural or semi-rural areas. The idea of community care has been around since the 1960's, but the nature of the care that is provided and the framework within which this care has been delivered has changed across time (Kirk and Glendinning 1998). Community care can be seen in terms of a move away from institutionalised care to community based care for people based in long term hospitals and as a move from institutional to community based care within the National Health Service, with an emphasis on Primary Care and the aim of reducing hospital based care levels. The growing crisis in the NHS in the 1970's, the costs of providing institutional care combined with a growing number of scandals relating to the quality of the care being provided within institutions led to an initial move away from large scale institutional care towards care in smaller, group homes within the community. This resulted in the closure, throughout the 1970's and 1980's of a large number of long stay hospitals for older people, people with learning disabilities, people with visual impairments and a whole range of other people with complex care needs. It was not until the arrival of the Conservative government in 1979 that the emphasis in social policy shifter away from care in the community towards care by the community – that is care provided by family, friends, neighbours and voluntary organisations (Parker 1990).

The policy of institutional closure and the promotion of community care can be seen in the recommendations put forward by the World Health Organisation in 1982:

> The strategy for attaining health for all by the year 2000 will fail unless we succeed in moving the pendulum back from the misuse of high technology, hospital based health services. Health is primarily a matter of self-care. If that approach is to permeate out society we must harness the support of individuals, families and health professions by inculcating a philosophy of primary health care. A more personal title, which reveals the foundations on which it must rest to succeed – the caring community – is community care. (1982: 48)

An increasing emphasis on the role of family carers can be seen from this point. The Conservative election win in 1979 led to a restructuring of the welfare state system which was driven by the new right. This led to a new language and rhetoric of welfare. Privatisation; decentralisation of services; quasi-markets and competition were the buzz words, and the state was left to regulate the financing of care for disabled people, people with mental illnesses, older people and other vulnerable groups (Wilson 2000). As Wilson points out, the boundaries of the state were being pushed back, and families and private care providers were increasingly being expected to provide care for these vulnerable groups. At the same time there was still a tension between residential and

community care. Throughout most of the 1980's it was easier to access residential care than get the money needed to stay in the community.

With mixed messages abounding and community care in disarray, the former chairman of Sainsburys, Sir Roy Griffiths, was brought in to review existing services and discover the way forward. The 1990 Community Care Act was the single most important policy document to emerge from the Griffiths Report in the 1990's. For the first time in the history of the welfare state there was a move away from universal provision towards the provision of a selective, targeted set of services (Wilson 2000). The focus was on reducing the amount being spent on private residential care, particularly for older people (Lewis and Glennerster 1996). At this point the change from care in the community to care by the community was reiterated and the role of family, friends and neighbours was re-emphasised. A corresponding change occurred within the statutory care agencies at this time with a focus on providing support for the family, friends and neighbours providing care rather than for the individuals who actually need the care. Although this is a very brief outline of the development of community care, it highlights the increasing use of family and friends as carers and opens the way for a more in-depth exploration of the literature on caring and informal care and the impact of this on the carers, the wider families and the people receiving the care. Before going further, however, it is important to note that the vast majority of dependent people have always been cared for informally outside institutions by family members, a fact which is often obscured by the focus on the impact of community care on informal care.

Caring

Throughout the last 40 years a considerable amount of research has been carried out, both as a direct result of the community care legislation and also preceding this, because of the increasing burden of care being taken on by informal carers. The research looks at the various aspects of caring, the impact that it has on the carer and their family in terms of health, quality of life and psycho-social well being, and at the financial implications, both on an individual carer level and at a National level and on the gender implications of informal care. In this section the wider literature on caring is reviewed before we focus in on the research that has been carried out exploring the particular impact of caring on parents caring for disabled children. We start by defining the concept of informal care and then move on to examine the type of people who provide informal care, the relationship between formal and informal care, the psycho-social impact of care on the carer, the financial implications of being a carer and the particular implications of being a carer of a disabled child. The experiences of families with juvenile Batten disease are then examined in the light of the wider trends within caring research.

The definition of informal care proposed by Twigg (1992) is used here, with informal care being seen as care that:

> "...normally takes place in the context of the family or marital relationships and is provided on an unpaid basis that draws on feelings of love, obligation and duty." (Twigg 1992: 2)

Four key aspects of informal care have been identified. Firstly, informal care takes place within the family environment with the majority of care, aside from within marital relationships, being provided by female relations (Phillips and Bernard 1995), very little care is provided either by the 'moral community', churches or ethnic groups (Abrams 1980) or by neighbours (Mays 1997). The second point is that informal care is unpaid (Kirk and Glendinning 1998), the third point is that it is based on the fact that the carer loves the person who they are caring for, the care is provided through love (Ungerson 1990). And the final point is that the likelihood of providing care increases with age, older people being more likely to be required to provide care than younger people (Mays 1997).

Gender also has a huge impact on the provision of informal care and a huge amount of research has been carried out looking at the links between gender and care. Figures from the 1990 General Household Survey suggest that 17% of women and 13% of men were providing care on a regular basis for a dependent person. Although the percentages of men and women providing care are similar, the fact that there are more women then men in Britain means that, in practice, 3.5 million women are providing care as opposed to 2.5 million men (Hillier and Scambler 1997). The Social Policy Research Unit (1994) elaborated on this to suggest that as many as 6.8 million people provide informal care in Britain, 1.7 million of whom provide more than 20 hours of care per week. Mays (1997) also suggests that not only are there more women providing informal care, but they are also far more likely to be providing

intimate personal care and physical assistance than their male counterparts. Green (1985) elaborates to suggest that women are more likely to provide care outside of the family home and to be the main carer. They are also more likely to spend more hours caring per week, with unmarried women between the ages of 45 and 64 being the most likely candidates for the role of informal carer, and are less likely to be receiving assistance.

There is widespread agreement on the 'burden' of care, and the stress that it places on the carers. A huge number of studies have been carried out across the world focusing on the difficulties and implications, for the carer, of providing care for a family member with a chronic condition. Family carers have been documented as suffering from stress and the disorganisation and disruption of family life, amongst other things (Loukissa 1995). Loukissa goes on to chart the change in the nature of research into family care for people with chronic mental illness. She notes the tendency to focus, throughout the 1960's and 1970's, on patient symptomatology and functioning levels, and highlights the move, in the 1980's to a focus on "caregiver's emotional strain, patient's behaviour and symptomatology, caregiver gender, and education, financial strain, and need for family education and support" (1995: 56). She suggests, further that in the 1990's there has been a move towards focusing on the relationship between the carer and the cared for, gender and levels of family education and support.

Whilst informal care can appear to be the cheap option for the statutory services, there is evidence that a wide array of costs may in actual fact be borne

by the carers themselves. These manifest themselves in the form of financial, physical and psychological pressures. The financial strain of caring is well documented. A study carried out by Henwood in 1990 found that just under half of all informal carers, whilst providing care for a minimum of twenty hours per week, were also holding down paid employment in order to support themselves and their families (Henwood 1990). Although financial support is available from government agencies for people involved in informal care work, payouts are difficult to get hold of and, where they are received, are often not significant enough to live on without additional financial support of some kind (Mays 1997).

The physical and psychological strain of caring has also been highlighted and, since 1948, respite care has been seen as the main way of providing carers with a break (Nolan and Grant 1992). A review of respite care carried out by Cotterill et al (1997), however suggests that the existing services may not be meeting the requirements of carers because of the difficulty of accessing appropriate respite services at the times when they are required. They suggest that the accessibility and flexibility of services need to be improved if they are to be of maximum use both to the carers and the service users. In addition, Cotterill et al (1997) suggest that the focus is now moving away from the provision of relief for carers and beginning to also incorporate the idea of providing a break for the service users as well. The benefits of respite care have been challenged by Cocks (2000) who, in an analysis of the role that disabled children play in the care that they receive, suggests that respite centres

can be seen simply as enforcing the difference and segregation that the children will be faced with throughout their lives.

> "The experience of childhood for disabled children who visit respite centres varies from their mainstream counterparts. This difference becomes apparent when considering the way in which the segregation of disabled children and their treatment as passive recipients of adult interventions takes place within a context of adulthood which is also segregated." (2000: 517-518)

She goes on to suggest that respite care aids in the construction of disabled children's childhoods in the model that society deems appropriate for them as disabled adults.

Studies have also been carried out looking at the impact of informal care on the people being cared for, suggesting that informal care, whilst being heralded as a positive development, is not always a positive experience for the people being cared for (Fox 2000). Goffman (1970) and de Swaan (1990), for example, suggested that patients could be both unprofessionally labelled and stigmatised within the home setting, whilst Graham (1979) and Dunlop (1986) have looked at the impact of the politics of domestic life on both the carers and the people being cared for. Building on these themes, research carried out in the United States of America looks not just at whether care is being provided informally by the family for family members who are chronically or acutely ill, but also at

how well this care is being provided. Schumacher et al (1998) suggest five criteria which need to be met if 'good' care is to be provided. These are:

1. Caregiving Mastery - having a positive view of one's abilities and skills around providing care.
2. Caregiver Self Efficacy – the carers views on their ability to positively cope with the physical and psychological problems of those they care for.
3. Caregiver Competence – self and professional perception of whether care is being provided adequately.
4. Caregiver Preparedness – how ready the carers feel that they are for the caring tasks that they will be undertaking.
5. Quality of Family Caregiving – this is the overall quality of the care provided by the family for their family member.

Schumacher et al suggest that the meeting of these criteria will ensure that care good quality is provided, and thus will take the strain of professional carers such as nurses. It is interesting to note, however, that the criteria are judged through the self perception of the carer and through the perception of the professionals. No account is taken of the opinions of the care receiver and whether they feel that the care that they are receiving is, or is not of good quality.

The final aspect of the literature on caring which needs to be mentioned in this context is that around the role of parents in the care of children with disabilities.

A considerable number of studies have been carried out looking at the role of parents in the care of their disabled children and the relationship between parents and professionals in organising and carrying out care (Case 2000; Murray 2000, Dale 1996) and the inequalities faced by families caring for children with a disability (Dowling and Dolan 2001). Research suggests that parents rarely have the expertise, knowledge or power which would allow them to become equal partners in decisions about the care needed for their child (Case 2000; Oliver and Barnes 1998; Dale 1996). Murray (2000) takes this a step further to suggest that it is the dominance of medicine and the medical model which places the emphasis on the idea of disability as the failure of the individual child which causes problems in the interaction between parents and professionals and will result in the disadvantage and oppression of the child (Murray 2000). Further to this, Dowling and Dolan use the social model to show that it is not just the child that is disadvantaged but that the inequalities are faced by the whole family trying to live in a society which is set up to disadvantage rather than equalise disabled people (Dowling and Dolan 2001). The impact of caring for a child with juvenile Batten disease was explored in the previous section in relation to the daily lives of families living with the condition. Particular aspects relating to the provision of informal care are reviewed in the next section.

Juvenile Batten Disease and Caring

In the previous chapter a whole section was dedicated to the specialist care needs of children and young people with juvenile Batten disease. A number of

points were also raised about the impact of providing care on the wider family. These related to practical issues around the provision of care, the emotional impact of caring for a child with a terminal illness and problems of maintaining the life of the family as a whole. The impact of caring for a child with juvenile Batten disease can be seen to mirror the broad themes apparent in the wider literature on caring reviewed in the previous section. Taking each of the themes in turn, the similarities are apparent. Before outlining the similarities between the experiences of carers of young people with juvenile Batten disease with those of the wider carer populations, it is useful to review the, limited, literature specifically on care in relation to juvenile Batten disease.

Although, as already outlined, very little research has been carried out looking at the social implications of juvenile Batten disease on the child and their family, five studies have been carried out looking at the particular care needs of the young people and their families. The one study looking specifically at the impact of care was conducted by Labbe (1996) in Australia, looking at the family functioning of care givers and the emotional states of primary care givers of children with Batten disease in comparison with the emotional states of primary care givers of chronically ill children with less severe illnesses. Primary care givers of children with Batten disease were found to be significantly more anxious and depressed, reported greater negative effects on their schedule and health, and perceived their families as less cohesive. The remaining social research that has been carried out looks more specifically at the social care needs of children and young adults with juvenile Batten disease. Coen de Jong (1996) identifies six key areas of care, incorporating the

accumulation and interpretation of information by the family; the impact of the emotional development of the child on their ability to cope; the need to make knowledge available to those involved in the day to day care of the child; and the need to provide emotional support for the families and carers. Shroigen (1993) has carried out research looking at the psychological impact of the disease on the children and young adults, and Von der Dunk and De Jong (1992) have identified the need to support carers through the terminal stages of the illness as well as the family and the child themselves. Scambler (1999) looked particularly at the social care needs of young adults with juvenile Batten disease and found, amongst other things, that the majority of parents are not happy with either the way they are given information, or the amount of information they were initially given about the disease and that carers, clinicians and therapists need to work in partnership with the family in order to provide the most appropriate and effective care.

Alongside the specific issues raised above, a number of themes were common to the experiences of families with juvenile Batten disease and the experiences identified within the wider literature. These include the financial, emotional and psychological impact of caring for a child with this condition. Although it was not a key theme to emerge from this study, a number of the families talked the financial costs of providing care in terms of the need for both parents to keep working or the strain placed on the family when the main carer, often the mother, was forced to give up work in order to provide the extensive care needed by their children. Gender differences were also consistent with the wider literature where it was noted that, in my sample, it was always the mother

who gave up work if one parent needed to provide full time care within the home. In addition to the financial costs, emotional costs were also mentioned, predominantly in relation to the terminal, degenerative nature of the disease and the fact that, as one mother explained, parents are going through the grieving process from the moment of diagnosis right through to the point where the child dies. The emotional strain on the families, watching their loved one lose all of their skills and faculties over a period of twenty years, is extreme and undeniable. The loss of verbal communication and facial expression were highlighted as two of the most difficult loses to face as a parent and carer. Emotional costs were also evident in the fact that half of the women who took part in the study are no longer with the father of the child with juvenile Batten disease, a fact described, by two of the mothers, as directly related to the stress of having to cope with the implications of the diagnosis.

Psychological costs were also highlighted in terms of the effect on the daily life of the family as a whole. This was illustrated in terms of a lack of spontaneity, the psychological difficulty of coping with the presence of a genetic disease within the family and the guilt of parents and non-effected siblings, the lack of time available to devote to the needs of other, non-effected, children and the pressure and feelings of guilt felt by parents when the difficulties of carrying out 'normal' family activities such as family holidays overcame them and they had to deal with the idea that other family members were missing out on the 'normal' family experience. All of these problems can be seen in terms of the psychological costs of caring for a family member with juvenile Batten disease

and would be likely to similarly effect families caring for children with other, similar genetic or profoundly disabling conditions.

Another area of informal care which was highlighted and is common to both families with juvenile Batten disease and to other carers was that of respite care. In spite of the view put forward by Cocks (2000) that respite care is damaging because it perpetuates the existing segregationist culture towards disabled people, respite care was identified as an extremely important aspect of care provision both for the young people and their family carers. This was the one area of care that was identified as lacking by the majority of the parents in my study. The provision of age appropriate respite care for adolescents and young adults is almost non-existent and the lack of adequate respite care provision, for young adults with juvenile Batten disease in particular, was highlighted as a key concern by the parents interviewed. This is in line with the findings of a recent report into local authority support for carers compiled by the Social Services Inspectorate (November 1998). The report found that:

> Short-term breaks and respite care services – services which are intended to provide particular benefit for carers – needed attention. While those that existed were welcomed and praised, more were wanted. Some respite services were felt to be unreliable or of low quality...(SSI, 1998: 6)

It is clear from the findings of the Social Services Inspectorate that this is a gap in care provision which effects a whole variety of people with a range of

different needs and is not simply limited to young people with juvenile Batten disease. The particular problems faced by the families in this study are due specifically to a lack of places and the minimal amount of time allocated to each family, the need for specialist care and the time to build a relationship with the young person, the scarcity of hospices and the considerable distances that families often have to travel in order to take up respite places, even where they are only for 24 hours, and the fact that, until the last two years, there were no hospices in England specifically for late teens and young adults.

The debates around parent/professional power struggles reflected in the literature were also present in the divergence of the views offered by parents and professionals when asked about the role of families in the care of young people with juvenile Batten disease. Whilst of the respondents - whether family members; carers; teachers; nurses or other medical professionals – emphasised the importance of the role of parents in care provision, there were differences in the opinions given as to whether parents were actively involved in care provided outside of the family home. The vast majority of the professional participants stressed the effective methods of communication already in place to ensure that were fully involved in the process of care. However, the majority of the parents interviewed felt either that their input was not as valued as they felt it ought to be, or that they had to fight to get the information that they wanted, or that they were not as informed about the day to day care of their child as they would like to be. In addition to this, all but one of the parents suggested that the amount of information and detail that they were given depended entirely on which particular member of staff they talked

to when they phoned. Thus, it was widely felt that, even where the school or unit had guidelines or policies to cover the sharing with information with parents, the amount of involvement that the parents had was dependent on the individual members of staff on duty at the time. The fact that there are such broad differences in the views on the levels of communication between parents and professionals is a clear indication that there are communication problems between the groups. The feelings of the families also suggest that, even where the caring is carried out in terms of decision making and emotional input rather than physical action, the effects of caring on parents of children with juvenile Batten disease are still considerable.

Self-Help

There is a growing literature on the impact of self-help groups, both within the chronic illness literature and within wider medical sociology, looking at power relations and challenges to orthodox medicine. In the following sections we look in more detail at the literature on self-help, its definition, functions and impact on wider society and the medical profession as a whole. We then examine these findings in relation to the setting up of the Batten Disease Family Association, and the impact that this self-help group has had, in the three years since its inception on the daily lives of families with juvenile Batten Disease, and at whether it fits in with the wider implications of the self-help movement.

There is some disagreement as to exactly what constitutes a self help group. In his analysis of the role of self-help groups, Kelleher (1994) outlines the key positions. The common description employed by Katz and Bender (1976) and Robinson and Henry (1977) is of a small group of like minded people getting together on a voluntary basis to provide mutual support. Whether self-help groups support or challenge the medical orthodoxy, however, is open for debate. Vincent (1992) states that the challenging or orthodoxy and politicising of the issues surrounding living with a chronic condition is a key role of groups of this kind. Kelleher (1994) suggests an alternative definition, incorporating some of these points and suggesting that:

> "The definition of self-help groups offered here, then, is that they are groups which place a value on experiential knowledge, thus implicitly challenging the authority of professional health care workers to define what it is to have a particular condition and how it should be managed." (1994: 111)

Another way in which to understand what constitutes a self-help group is to look at the types of activities and functions which they undertake. It is useful to start, here, with the classification provided by Katz and Bender (1976) who classified self-help groups by whether they were inner or outer focused groups. 'Inner-focused' groups were seen to provide support for their members and a forum through which they can share experiences, while 'outer-focused' groups functioned more as pressure groups raising the profile of the condition either

amongst professionals or the general public. Kelleher suggests, however, that whilst this is a good starting point, many groups seek to perform both functions, whilst others perform different functions altogether. Williams (1989), in a study of the National Ankylosing Spondylitis group, suggests that the people controlling the group will also effect the nature of the group, with professional led groups being less likely to challenge the existing medical orthodoxy, although they may campaign for greater awareness and so forth. Groups that originate from, and are led by, sufferers, however, are more likely to feel able to criticise orthodox medicine where they feel such criticism is deserved. Gabe (1995) uses the example of a local group which was set up to support people addicted to tranquillisers to illustrate this point. He suggests that, whilst the groups originally set out to provide a forum where members could share experiences and support one another, they soon turned to campaigning for greater awareness amongst medical professionals, suggesting that more information should be given about potential problems with the treatment before tranquillisers were prescribed. This group turned from an 'inner-focused' to an 'outer-focused' group and set out to challenge medical practice.

A third function of self-help groups is highlighted by Kelleher (1990, 1991) who looked at the work of the British Diabetic Association, the main aim of which, at the time of conception, was to raise money to support research in the area of diabetes. Finally, it seems that self-help groups may also be set up to challenge the dominant view that conditions, and particularly chronic conditions need to be seen predominantly from a medical perspective (Kelleher 1994). A number of studies have been carried out looking at this aspect of the

groups, and suggesting that a key focus may be to turn attention away from medically focused definitions, and to refocus on the day to day experiences of the condition, examining what the condition is like for the people who live with it on a daily basis, and at the narratives that they construct around their experiences (Arntson and Droge 1987, Bury 1988, Williams 1989). It seems clear from this that self-help groups are set up to perform a range of functions. The groups may be led by professionals or by sufferers, or, in the case of children with particular conditions or disabilities, by the families of sufferers. The groups may seek to provide supportive platforms through which people can share experiences, or they might seek to provide information, or challenge orthodox opinions, to redefine the parameters within which a particular disease is understood, to raise the profile of conditions, either amongst professionals or the lay population, or to raise money for research. Often, however, more than one of these tasks is undertaken.

Having established a general definition of self-help groups and the range of activities in which they participate, the literature on self-help goes on to explore the role of self-help groups in contemporary culture and how and why they have come to be so prolific. Kelleher (1994) suggests four possible reasons for the proliferation, from the late 1970's onwards, of the self-help group. The first possible reason is offered by writers looking at the state of contemporary society and suggesting that it has lost its sense of purpose and cohesion. Giddens (1991) and Bauman (1992), for example, suggest that people feel that they are unable to control the society in which they live, they are wandering aimlessly rather than forging ahead purposefully guided by knowledge of right

and wrong. This point is elaborated on by Lasch (1980) and Sennett (1977) who highlight the propensity of people, in this uncertain situation to turn in on themselves and seek inner satisfaction rather than seeking active involvement in society as a whole. This is one suggested reason for the growth in self-help groups, where people look to their own, individualistic problems and needs rather than to those of wider society as a whole (Kelleher 1994). In a similar vein, the second possibility is that a lack of control over the wider environment, combined with a focus on health and being able to control ones health (Glassner 1989), has led to a growth of groups supporting the individual in their quest for health. The proliferation of self-help groups may then be attributable both to increasing individualisation and emergence of a risk society (Beck 1992), supported by the Conservative government of the 1980's which promoted the individualistic approach as a cost cutting measure (Kelleher 1994). The third possible explanation for the growth in self-help groups proposed by Kelleher is that people with chronic long standing conditions need support to cope with the everyday, mundane aspects of living with chronic conditions, things like managing stigma and developing coping strategies, which are not covered by the health service. Thus they are forced to seek this support from elsewhere, and turn to self-help and the support of other people in the same or similar situations.

The final possible explanation that has been offered is that self-help groups have developed as a challenge to the dominance of the medical profession over issues of health. Calnan and Williams suggest that the modern patient, as an articulate consumer of medical services, rather than a passive recipient, poses

challenges to the traditional doctor/patient power relations (Calnan and Williams 1996). This perceived change in the relationship of the lay population with medicine has been explored by a number of people. Elston (1991), for example, suggests that a less deferential attitude towards expert knowledge, along with increased knowledge and access to information amongst the lay population has had an impact on the expectations and behaviour of patients. Kohler Riessman (1989), however, suggests, whilst looking specifically at women, that in retrospect, patients have always been active, although levels of interaction and involvement have depended on the social class of the patient. Challenges to the power of medicine have been mounted in a number of areas including those of chronic illness (Silverman 1987, Anderson and Bury 1988) and disability (Oliver 1990, Swain 1993, Bickenback 1999) where it is widely felt that medicine has very little to offer in the way of day to day support or understanding of the true impact of either chronic illness or disability on the everyday lives of the people living with them.

The emergence of the self-help movement is heralded as one way in which the lay population are challenging the dominance of medicine and prioritising the knowledge that they themselves hold (Kelleher 1994). Kelleher sees this as an enaction of a type of "lay resistance" or challenge to the dominance of medical, scientific and technological dominance of this arena. He suggests, further that medical dominance is being challenged both from above, through the increased power of managers, and from below, through the increasing numbers of people, particularly those with chronic illness who feel that they cannot be helped by medicine, turning to each other for help rather than to the medical profession.

The growth of self help can be seen to originate from the development of groups set up to support people with a whole range of conditions, disease and disabilities in the 1970's. Figures from the USA suggest that as many as 20 million people were involved in self-help groups in the USA by the end of the seventies (Arntson and Droge 1987). Similar trends were found in Europe (Kickbusch and Hatch 1983) and in England (Vincent 1992), although exact figures for the number of groups in existence are hard to determine due to the specialised nature of many of the groups and the limited life spans of others.

Changes in the way in which the lay populace views medicine are also explored by Cornwell (1984) who proposes that the relationship between lay people and medicine can be explored on different levels. He suggests that 'rationalisation', or the progression and development of lay people's perspectives on medicine, can be examined using a Habermasian distinction between 'traditional' (based on religion, philosophy and morality) and 'modern' (based on science and technology) legitimation. He sees a move towards 'modern' legitimation, and a knowledge based on science and technology, empirical facts and analytical knowledge rather than 'traditional' legitimation as occurring unevenly across society. Further, he sees this move as occurring at two levels, both at the level of the cultural and also at the level of the individual or sub-cultural. Cornwell suggests that this is a useful way of conceptualising the change in lay attitudes towards medicine.

> It states the dominant tendency in our culture, which is towards modern scientific and technical forms of legitimation, without

> implying that the process will necessarily be carried through everywhere, and in all social groups, at the same pace or at the same time. The rate of progress of medicalisation depends upon the state of readiness of the sub-culture and individuals within the sub-cultures to allow it to take place, and on their state of awareness and knowledge of scientific achievements. (Cornwell 1984: 120)

If this is the case, the parents of children with juvenile Batten disease who have formed the Batten Disease Family Association are both part of the movement towards a 'modern' legitimation of medicine, realising the central importance of medical development to their chances of treatment or a cure, and also part of the challenge to medicine posed by self-help groups through the acknowledgement that medicine can only provide part of what is needed for families and that often the families know more that the professionals.

This begs the question of the exact nature of self-help groups, bringing us in a full circle from the initial discussion over the definition and functions of a self-help group. Kelleher (1994) suggests that Habermas's 'Theory of Communicative Action' (1987) and work on the nature of new social movements (1981) can be used to make the argument that self-help groups are a form of new social movement. Habermas argues that there are 'differentiated mechanisms of instrumental-cognitive rationality' which help to maintain and develop the social world. Expert systems are one of these mechanisms. He also argues that there is a life-world which is made up of the general population

and which is based largely on traditional values. Kelleher (1994) suggests that self-help groups can be found within the life-world acting as a challenge to the dominance of medicine. In the words of Kelleher:

> Self-help groups can be seen as part of a new social movement which is resisting the domination of the life-world by expert systems, in this case the expert system of medicine. (1994: 113).

Habermas suggests that the life-world has become separated from the expert systems and is seeking to challenge the relevance and dominance of the expert system to itself through new social movements. Further he proposes that the life-world has, in effect, been colonised by the expert systems which determine much of what they do, how they act and the framework within which they understand their own lives (1981), and in this instance, their health. This being the case, self-help groups can be seen as a challenge to the colonisation of experiences of chronic illness by the expert system of medicine and/ or as a way of re-engaging the life-world with the expert system. Again Kelleher elucidates the impact of the self-help movement in these terms:

> Self-help groups, then, are part of a public sphere where life-world concerns can be discussed in the language of the life-world. In providing this opportunity they can be seen as part of a social movement which is resisting change, withstanding the drive to understand human experiences in ways that deny the value of

> knowledge constructed intersubjectively by means of communicative action. (1994: 115).

Self-help groups, thus are an important way of maintaining the importance of experience and the identity of the patient as more than a medical case. They can be seen in terms of a challenge to medical power whilst not necessarily denying the important role that medicine can and does play. In the next section we explore the emergence of the Batten Disease Family Association, and the idea of group identity.

The Emergence of a Group Identity

It is possible to suggest that a form of group identity was developed even preceding the formation of the Batten Disease Family Association. Drawing on the work of Calhoun, Bourdieu and Habermas, amongst others, the suggestion is made that the families of children with juvenile Batten disease were forming their own identity with common experiences and goals, in spite of the fact that they may never have met or even communicated with one another, merely through the knowledge of the existence of one another, before an official forum was established.

Calhoun (1994) uses the work of Kluckhohn and Murray (1948) as the basis for his understanding of group identity. They categorise all people by saying that:

Every man is in certain respects

a) like all other men

b) like some other men

c) like no other men (1994, p176)

It is the second of these statements 'like some other men' that leads to the emergence of a group feeling or identity. This idea was looked at in more detail by Bourdieu in his work on 'habitus'. 'Habitus' is shared traits of feeling, conduct and taste which separate one group from another, or more importantly, draw groups closer to each other. Bourdieu suggests that this is largely an unconscious sharing, whereby the members of the group do not consciously realise that they are acting in a way peculiar to their group. Calhoun takes Bourdieu's idea of habitus and suggests that it is linked to the modern concept of identity, the only difference being that the concept of identity suggests a more conscious awareness of the differences that the group shares. Calhoun states that:

> "'identity' implies a higher level of conscious awareness by members of a group, some degree of reflection and articulation, some positive or negative emotional feelings towards the characteristics which members of a group perceive themselves as sharing and in which they perceive themselves as differing from other groups."

He goes on to suggest that in modern society these group identities are multi layered and every person will be in a number of different groups which they will belong to with more or less conviction or reason.

Following this, the argument can be made that being part of a family that is affected by juvenile Batten disease is the basis upon which a new group identity emerged. Add to this the fact that the disease goes against all that would normally be expected in the way of development and the reasoning becomes clear: "the child you've seen growing up normally all of a sudden has this illness that you know is not going to get any better, it's going to get worse.". There is a feeling that, in the words of one mother, "nobody understands better than another parent". Among the families with younger children there was a considerable amount of communication and information sharing, along with an acknowledgment and welcoming of the fact that there were other people out there with the same problems who really knew what it was like. "You find out things from other parents", "Information is best given to you by other parents who've been through it", "so called professionals can only read what they know out of a text book. Parents can actually say they've lived it, breathed it, experienced it." One mother talked of the support that she receives from another mother:

> "I've got a couple of mums that have children with Battens that I ring up if I've had a very bad day or if I've had a very good day, you know, I ring up and say guess what we've just found out about

this, or I ring up and say [my daughter] had a really bad day today and I'm really piddled off."

The formation of the Batten Disease Family Association can be seen as the formalisation of these links and the group identity that was emerging even before the BDFA was physically formed. The characteristics of a group identity are now clearly visible. The families share feelings of alienation and the struggle to keep on an even keel when they are moving in the opposite direction to the rest of the population. They share conduct in so far as they are all constantly fighting with the Local Authorities and Social Services to give them what they need and in that they are always fighting to keep their children as involved in society as possible. They also share the need to come to terms with a disease that is horrific in its ramifications for the family as a whole.

The emergence of a group identity brought with it an informal self-help group in the sense that the families were sharing experiences, knowledge and ideas with each other and helping one another to cope with the implications of the disease. This provided the basis upon which the formal structure of the Batten Disease Family Association (BDFA) was built, and through which the group has evolved into something which is more permanent, enduring and formal. It is the development of the BDFA that is now explored in some detail with recourse to the empirical data collected throughout the conception, formalisation and first two years of the life of the group.

The Batten Disease Family Association

In order to look at the impact of the Batten Disease Family Association, the roles that it seeks to perform, the impact that it has on its members, and whether or not it fits in with the wider literature on challenging medical orthodoxy and new social movements, it is useful to outline the development of the movement. This is done through a detailed examination of the setting up, legitimation and running of the group, from its inception right through to its gaining legitimate status as a charity. In the course of my data collection I was able to carry out a two year participant observation of the setting up of the Batten Disease Family Association. There are a number of key stages that the group moved through which are explored in more detail below.

The Initial Meeting and Display of Interest

All of the parents that took part in the initial interviews for this study knew of, and had been in contact with, at least two other families. The parents of the younger children had had a considerable amount of contact with other families to the extent that they expressed a desire for a more formal group where they could get together and support one another. An initial meeting was organised by seeAbility, a charity that was involved in research looking at the specific care needs of young adults with juvenile Batten disease. The meeting was held in November 1998, and all of the families that were known about by the organisers were invited. In all, 15 families attended the meeting representing the two most common forms of Batten disease, juvenile Batten disease and late infantile Batten disease. A further ten families expressed an interest but were unable to attend the meeting. The Development Officer from the charity

'Contact a Family' was also invited and gave a formal presentation on how to set up a family association, setting aims and objectives and formalising a constitution and charitable status.

The woman from 'Contact a Family' talked about various issues around setting up a support group for a rare condition such as Batten disease. She split her talk into four key points:

1. Why set up a group?
2. Key times when the group can be of most benefit.
3. Benefits.
4. Considerations.

She suggested that a group could be set up because of the satisfaction of being able to actively do something useful; to provide support for newly diagnosed families; as a way of raising awareness of the condition as a whole; as a way of sharing information between families and the medical profession; and to provide friendship and a forum for socialising. Moving on she highlighted the key times when the group could be of most use, focusing on diagnosis; times when new services are required (because there is no continuity of services); through the education system where great changes in the circumstances of the child will occur; at the point of transition from child to adult services; at times of crisis and at the point of bereavement. She suggested that all of these points were ones where the group could be of particular use. Linda then went on to explore the benefits of setting up a group of this kind. She talked about the

relief of the isolation of being a family with a rare condition and the knowledge that there are other families out there in similar circumstances. She also talked of the importance of providing information and of information exchange and sharing things that have and have not worked. Other benefits included the knowledge that you can offload problems, stresses and worries in a safe environment with people who would understand and reciprocate; the provision of hope; and the opportunity for time out and to meet up with similar people at least once a year.

The final point that she focused on was the various things that need to be decided on before a group of this kind can be set up. Ten areas were highlighted for consideration:

1. The need to decide what the group is going to offer to its members.
2. The need to take on board the expectations of the families and look at whether they can be met.
3. The need to be realistic. The group will be run by family members and must fit in with the pattern of the families.
4. The need to allow for growth and to develop a plan of action for the next 12 months and meet the action points.
5. The need to network and let other groups know of your existence.
6. Practicalities of who is going to do what.

7. Financial implications of running a groups of this kind – being a registered charity can help in raising money, even the relatively small amounts needed for telephone work and printing.
8. The need to take the group forward, and set up a committee to meet three or four times a year either face to face or via telephone conferencing.
9. The need to decide the aims of the group and stick to them.
10. A number of questions were raised about the setting up of the group.

Q What about groups who do not want you to set up and make it difficult?

A There is room for every group to exist.

Q What about links with the American group?

A Practically it is difficult to become part of another group. Must remember the rest of Europe and move to set up groups across Europe.

Q Are there advantages to having very large groups – globally?

A Experiences may be different and it is difficult to manage and make sure large groups are democratic and meet the needs of the members.

The parents were then spilt into groups and asked to discuss what they wanted from the group. The families decided on a steering group consisting of five parents. They also received offers of initial funding from representatives of two other charities, the 'Daniels Charitable Trust' and 'seeAbility'. A list of the names, and contact details of all of the interested families was also compiled. Short term objectives were set at:

- Compiling a list of people willing to help.
- Networking and letting other groups know of their existence.
- Compiling the first newsletter – maybe as a report of the day.
- Deciding on the name for the group.
- Identifying a medical advisory board.
- Setting up a website.
- Sharing the experiences of the families involved so far.

Finally there was some discussion regarding the name of the group. It was agreed that the steering group would circulate ideas – initial thoughts were: Batten's Disease Association; Association for Batten's Disease; Batten's Family Association; Batten's Disease Family Association.

Agreeing Aims and Objectives

At the initial meeting a questionnaire was handed around to all of the people in attendance. Questionnaires were also sent to all of the families that we had contact with to date. The aim of the short, single page, questionnaire was to

establish which aspects of the group the families felt were most important and what they would most like to see the group providing in the future. Altogether 30 questionnaires were sent out and we received 20 completed questionnaires. The responses to the questionnaire are shown in table 7 below:

Even from the response to this initial questionnaire the difficulties in setting up a group of this kind can be seen. There was little agreement on which aspect of the group should be most important. The results of the questionnaire do suggest, however, that a range of support services need to be provided to meet the needs of all of the members of the group (See Table 7 below). The results of the questionnaire were taken along to the first steering group meeting which was held on 6th February 1999. At the meeting the name 'Batten Disease Family Association' was agreed and a mission statement was agreed, to read as follows:

> "To provide a supportive, informative, networking organisation for families, carers and professionals giving care to children and adults with Batten's Disease."

A press release was also complied to be circulated around the existing groups to alert them to the existence of the new batten disease group. In addition to this the positions of the parents who had agreed to become steering group and committee members were agreed upon; the need for a web site and database of members were agreed; the content of the first newsletter were suggested and agreed; and the matter of a logo was discussed and opened for suggestions.

Table 7

Results of the BDA Questionnaire	
Questionnaire Responses	20 questionnaires were received - 10 from parents attending the meeting and 10 from parents not attending the meeting.
Type of Batten's Disease	5 responses were from families with late infantile Batten's disease and 15 responses were from families with juvenile batten's disease.
Linking Families in Similar Circumstances	4 rated this aspect of the group as most important. 3 rated it second, 4 rated it third, 3 rated it fourth, 3 rated it fifth, 3 rated it sixth and no one rated it seventh.
Sympathetic Listening Ear for Newly Diagnosed Families	6 rated this aspect of the group as most important. 6 rated it second, 2 rated it third, no one rated it fifth or sixth and 1 rated it as seventh in order of priority.
User Friendly Information on Batten's Disease	4 rated this aspect of the group as most important. 5 rated it second, 4 rated it third, 3 rated it fourth, 3 rated it fifth, no one rated it sixth and 1 rated it seventh.
Additional Information	No one rated this aspect of the group as most important. 1 rated it second, 1 rated it third, 2 rated it fourth, 3 rated it fifth, 7 rated it sixth and 5 rated it seventh.
Regular Newsletter	2 rated this aspect of the group as most important. 1 rated it second, 2 rated it third, 4 rated it fourth, 3 rated it fifth, 4 rated it sixth and 4 rated it seventh.
Information on Current Research	4 rated this aspect of the group as most important. 6 rated it second, 3 rated it third, 1 rated it fourth, 5 rated it fifth, no one rated it sixth and 1 rated it seventh.
Occasional National Meetings	No one rated this aspect of the group as most important. 3 rated it second, 3 rated it third, 1 rated it fourth, 2 rated it fifth, 4 rated it sixth and 5 rated it seventh.
Other	Other aspects mentioned included forming a partnership with the researchers towards finding a treatment, equipment exchange and advice and fundraising for the group.
Committee Member	3 people indicated interest in being on the committee.
Linking With Other Families	10 people said they would like to link with other families.
Compiling Batten's Info	2 people said they would collect and collate information.
Working on a Newsletter	4 people said they would be willing to work on a newsletter.
Helping Organise Meetings	5 people said they would be willing to help organise meetings.

The organisation of two seminars was also discussed, the first to be held in June and aimed predominantly at professionals and the second to be held in October and aimed at families. The need for a constitution was also agreed upon.

The objectives of the Batten Disease family Association, as laid out in the constitution (adopted formally on 13th April 2000), are as follows:

1. Preserve and protect the health and promote the welfare of persons affected by all types of Neuronal Ceroid Lipofuscinosis (NCL) commonly known as Batten disease.
2. To advance the education of the medical profession and the general public on the subject of Batten disease and its implications for the family.
3. To promoted research into the management of Batten disease and to publish the useful results thereof and to support organisations promoting research into Batten disease.

A further 24 powers were laid out relating to these four objectives. They are as follows:

1. Create a positive network of parents, families and professionals in order to share ideas, experiences and give emotional support.

2. Recognise the emotional needs of children affected by Batten disease and to encourage the recognition of the depth of feelings experienced by their families and provide support.
3. Increase the understanding of the relationship between the physical effects of Batten disease and learning development; and to aid the development of beneficial therapies and learning programmes.
4. Help parents obtain the best education and facilities for their children.
5. Support the parents in their right to be kept informed about the likely progression of their child's condition and more importantly the outcomes of treatments offered at various stages.
6. Promote exercise and physical endeavours as a beneficial therapy.
7. Aid the development and understanding of the possible benefits of dietary control in children with Batten disease including issues surrounding alternative feeding techniques and of alternative forms of medicine.
8. Help and support parents in all their negotiations for financial aid and other services.
9. Promote and support all activities leading to an improved quality of life.
10. Employ and pay and person or persons (who shall not be members of the executive committee) to supervise, organise

and carry on the work of the Association and make all reasonable and necessary provision for the payment of pensions and superannuation to or on behalf of employees and their widows and other dependants.

11. Bring together in conference representatives of voluntary organisations, Government Departments, the medical profession, statutory authorities and individuals.
12. Promote and carry out or assist in promoting and carrying out research, surveys and investigations and publish the useful results thereof.
13. Arrange and provide for or join in arranging and providing for exhibitions, meetings, lectures, classes, seminars and training courses.
14. Collect and disseminate information on all matters affecting the said objects and exchange such information with other bodies having similar objects whether in this country or overseas.
15. Undertake, execute, manage or assist any charitable trusts which may lawfully be undertaken, executed, managed or assisted by the Association.
16. Cause to be written and printed or otherwise reproduced and circulated, gratuitously or otherwise, such papers, books, periodicals, pamphlets or other documents or films or recorded tapes (whether audio or video or both) as shall further the said objects.

17. Purchase, take on lease or in exchange, hire or otherwise acquire any property and any rights and privileges necessary for the promotion of the said objects and construct, maintain
13. Increase the understanding of the relationship between the the Association.
18. Make regulations for any property which may be so required.
19. Subject to such consents as may be required by law, sell, let, mortgage, dispose of or turn to account all or any of the property or assets of the Association.
20. Subject to such consents as may be required by law, borrow or raise money for the said objects and accept gifts on such terms and on such security as shall be deemed to be necessary.
21. Raise funds and invite and receive contributions from any person or persons whatsoever by way of subscriptions otherwise PROVIDED THAT the Association shall not undertake permanent trading activities in raising funds for the said objects.
22. Invest the money of the association not immediately required for the said objects in or upon such investments, securities or property as may be thought fit, subject nevertheless to such conditions (if any) as may for the time being be imposed or required by law.
23. Do all such other lawful things as are necessary for the attainment of the said objects.

24. Establish where necessary local branches (whether autonomous or not).

As can be seen from the points above, the aims and objectives of the group had to be worded in such a way as to be acceptable legally when charitable status was achieved. The aims and objectives can be seen to reflect the key points raised by Kelleher (1990, 1991) in his analysis of the role of self-help groups. The Batten Disease Family Association is both inner-focused - in terms of seeking to provide support and information and experiential exchange amongst the families – and outer-focused – attempting to raise the profile of the conditions and experiences of the children and their families. The group also seeks to fund research into both medical and social aspects of the disease and to raise the profile of the disease by participating in national political action around raising awareness of genetic and metabolic diseases.

There is one aspect of the literature on the role of self-help groups that does not fit in with the aims and objectives of the Batten Disease Family Association, however. This concerns the challenge mounted by self-help to expert systems by the Lifeworld, as suggested by Habermas (1994), focusing on the support that can be given by 'ordinary' people, rather than that offered by 'experts'. As suggested earlier, whilst the BDFA may be part of the challenge to medicine posed by self-help groups through the acknowledgement that medicine can only provide part of what is needed for families and that often the families know more that the professionals, the group are also part of the movement towards a 'modern' legitimation of medicine, realising the central importance of medical

development to their chances of treatment or a cure, and thus adopting a position that puts them at odds with the majority of the sociological literature on chronic illness explored in the next chapter.

Reactions from Other Groups

The political nature of setting up a group of this kind was also experienced as the BDFA took form. Throughout the course of the months surrounding the setting up of the Batten Disease Family Association correspondence was entered into with a number of existing charities which seek to provide support for people with Batten disease. Although most of the feedback was positive we also received negative (or lukewarm) responses from two existing organisations who expressed reservations about the need for a new group. One such response was received from an existing umbrella organisation who reiterated the support that they had provided to families with Batten disease and seemed to think that the new group was unnecessary. The other less than positive response was received from the Batten Disease Support Association, run by a woman in Scotland who suggested that there was already a national group and a new one was not necessary. Both of these correspondences were forwarded to the parents of the steering group who felt that the umbrella group, although supportive and useful, was not adapted specifically to their needs, and that the other 'national group' was not providing them with the support they needed. None of the parents were members of the group - although some had heard and/or attended a conference organised by them in June 1997.

The majority of the responses and letters received during the long process of setting up the group were extremely positive and supportive, however. 'Seeability' were instrumental in setting up the group, providing support, financial and administrative support and the use of a purpose built location for meetings and conferences. 'Contact a Family' provided both practical assistance and support and the Batten Disease Support and Research Association – the American equivalent of the BDFA, although much larger – provided support and information and active interest in the development of the group. In addition the group of researchers based at University College London provided support and information and ongoing updates on the state of current research. The vast majority of parents and professionals were extremely positive and supportive of the setting up of the group.

The BDFA in Action

The provision of a regular newsletter was on of the key aims of the group at the time of its creation. The first three editions of the Batten Disease Family Association Newsletter can be found in Appendix One. The newsletters provide information about the activities of the group; reports on conferences attended or run by members; updates on the latest bio-medical research and daily-life stories sent in by the families of the affected children. They also contain membership details and are one of the most important aspects of the group's work. The other key actions of the BDFA so far are the successful organisation of three national conferences and playing a pivotal role in one international conference and providing £10 000 (raised by a variety of fund-raising events) for continued research into the study of mice developed with the

mutant gene thought to be related to late-infantile Batten disease, at the Department of Neuropathology, Institute of Psychiatry, King's College London.

By the time of the third steering group meeting, held on 22nd May 1999 the first draft on an information leaflet about the group had been produced, the forms for an application for charitable status had been completed and the first draft newsletter was complete and agreed upon. The BDFA was formally set up in July 1999, the Constitution was formally agreed on 13th April 2000 and, after a long wait, charitable status was awarded to the Batten Disease Family Association on Monday 12th February 2001. The Batten Disease Support Association is flourishing and looking to raise more money for research at the current time. They are also providing one to one support for all newly diagnosed families brought to their attention and have leaflets in all of the key centres coming into contact with families with Batten disease. A core group of parents have put a huge amount of work, time and effort into running the group, providing support and information, compiling and editing the newsletter, maintaining the database and financial records and all of the other tasks necessary to keep the group running smoothly.

Summary

This chapter explores the literature around caring and self-help and makes links between the experiences of families with juvenile Batten disease and of those with other conditions. The previous chapter outlined some of the ways in

which family carers of young people with juvenile Batten disease are more than simply carers and can be seen to experience the condition. This chapter extended this by comparing the experiences of the families with the wider literature and suggesting that, not only do families of young people with juvenile Batten disease experience the condition, they also face all of the other dilemmas and stresses facing the multitude of people providing informal care. In addition, the literature around self-help and the role of support groups was outlined before a detailed exploration of the establishment and legitimation of the Batten Disease Support Association with information gained throughout the course of a two and a half years participant observation of the setting up of the group. The nature of the BDFA as both an inner-facing and outer-facing group, which provides support for members as well as raising the profile amongst professionals and the wider population was established, along with their desire to provide financial support for bio-medical and social research and participate in political action to raise the profile of the disease. The final point to reiterate is the nature of the BDFA as a group which challenges the dominance of bio-medicine through an understanding of the need for support and information sharing amongst families, whilst at the same time acknowledging the central importance of bio-medicine and research in forwarding the cause if Batten disease and carrying on the ultimate search for a cure. In the next chapter this dual role is revisited in relation to the traditional sociological approach to chronic illness as laid out in chapter four.

7 Juvenile Batten Disease and Changing Experiences and Understandings of Chronic Illness

This chapter explores the experiences of families with juvenile Batten disease in relation to the wider literature on the experiences of people with a whole variety of more common chronic illnesses. Common experiences are examined through an exploration of the key themes that emerge through the sociological literature on chronic illness – uncertainty, stigma, family relations, biographical disruption and the reconstitution of self, managing the medical regimen and information, awareness and sharing - in an attempt to answer the third research question posed in the thesis: 'Do the experiences of families living with juvenile Batten disease reflect the wider experiences of people living with more common chronic illnesses?' This leads to two further questions, as highlighted in the introduction:

- Does the nature of juvenile Batten's disease fit in with the challenge to bio-medicine inherent in the sociology of chronic illness?

- Are arguments about the social construction of chronic illness relevant to the experiences of people with juvenile Batten's disease?

This chapter starts with an exploration of the extent to which the experiences of families with juvenile Batten disease fit in with the common themes emerging through the literature on chronic illness. From there the extent to which juvenile Batten disease fits in with the challenge to the dominance of bio-medicine is explored along with the relevance of social constructionist arguments around the creation of chronic illness. The last part of the chapter relates the key tenets inherent in the sociological literature around chronic illness to the ten themes identified in the introduction and expanded on through the bio-medical facts of juvenile Batten disease, highlighting any areas in which there may be discrepancies.

Juvenile Batten Disease as a Chronic Illness

The rarity of juvenile Batten disease raises interesting issues when confronted with a chronic illness literature developed almost entirely from the experiences of people with more common forms of chronic illness. The low incidence of juvenile Batten disease (affecting approximately 75 people in the United Kingdom at any one time), in common with many other rare chronic illnesses, means that any studies will, by necessity, have small sample sizes, thus limiting the validity and generalisability of their findings (Burr, 1985). This raises further issues about how well conclusions about one rare illness can be generalised to other illnesses (Burr 1985). Although, as argued in chapter four,

the data collection techniques utilised within this study give an overview of the whole picture of juvenile Batten disease within England, there is still very little additional information specifically on juvenile Batten disease, and so, much information has to be gained from external sources. Taking this on board, there are a number of ways in which the literature on, and experiences of, people with juvenile Batten disease have similarities with the broad themes raised through the wider literature on chronic illness.

Uncertainty

As suggested in chapter two, uncertainty is a common theme which appears repeatedly in the literature on experiences of people living with chronic illnesses. Uncertainty may related no just to an uncertain aetiology, but also to uncertainty surrounding the disease trajectory and the short and long impact that it will have on the life of the effected person (Turk 1979). This uncertainty has been documented in research carried out looking at the experiences of people with rheumatoid arthritis (Anderson 1989), Parkinson's Disease (Pinder 1990), Multiple Sclerosis (Robinson 1988) and HIV and AIDS (Aggleton and Thomas), amongst others. As illustrated in chapter five, where the experiences of the families were explored in detail, uncertainty plays a prominent part in the experience of caring for someone with juvenile Batten disease, and living with juvenile Batten disease.

From the onset of symptoms through to diagnosis is a passage that can incorporate a number of years, tests, specialists and misdiagnoses, throughout

which there is the uncertainty of not knowing what is happening or why (Scambler, 1999). Even when the diagnosis is finally received uncertainty plays a prominent role in the experiences of the family. The basic symptoms are the same for each child, however, the age at which each symptom develops and the severity of the symptom varies from person to person. Thus, the disease is different for every person who has it and so there is the uncertainty of not knowing what is going to happen next and when the next loss may occur (Schroijen, 1993). For the parents the uncertainty comes from a general knowledge of the disease course without any solid information on the timescale and severity of each phase. For the children the uncertainty can be seen to stem from that fact that they are unaware of the implications of their illness or its inevitable outcome (Scambler, 1999), as illustrated through the fact that none of the parents interviewed had chosen to tell their child either the diagnosis that they had been given or the implications of it. This is the first way in which the experiences of families with juvenile Batten disease clearly share common ground with the experiences of people with more common chronic illnesses.

Family Relations

The implications of chronic illness on the family as a whole, as well as on the effected person themselves, is well documented. Issues identified range from the role of the family in the care of the affected person (Smith 1979, Anderson 1998) through to the impact on the finances of the family (Conover 1973; Whitehead 1987). The impact of juvenile Batten disease on family relations can be seen throughout the literature on the disease (Scambler, 1999; Schroijen,

1993; Labbe, 1996). As chapter five showed, the disease has a huge impact on the maintenance and breakdown of normal family roles, and impacts on the lives of unaffected siblings and the wider family as well as the main parent carer/s. Parents are required to cope with the diagnosis of a terminal condition and the loss of the hopes and dreams that they would have had for their child. There is evidence that some marriages cannot take the strain (half of the mothers interviewed through the course of the study were no longer with the father of the affected child), siblings have less attention, career choices (particularly of mothers) are often adapted to the caring role and there are financial implications of providing care and equipment, as well as adaptations to the family home, which are exacerbated by limitations placed on occupational choices due to the burden of care (Scambler, 1999). This links to the documented need to provide support for the family of the child as well as the child themselves, tying in with issues identified within the chronic illness literature around family involvement in the disease processes.

The stress of caring for a child with the knowledge that their condition is terminal, over such an extended period of time, has also been identified as a problem and linked to increased levels of depression and anxiety (Labbe, 1996). As illustrated in chapter six, there are a range of issues effecting the choice and accessibility of care, both within the family home and in alternative locations, which also effect the family. Where care is provided within the home major adaptations to the environment are needed, and a considerable amount of parental time will be spent in providing increasingly technical, biomedical care. Where care is provided in an alternative location, families

experience feelings of guilt and dislocation as well as the loss of the parental role, and often lose a considerable amount of time through travelling – often extensive – distances to make regular visits to their child.

In addition to the more general stresses that would be faced by many families of people with chronic illnesses, the genetic nature of the disease brings a whole range of additional problems. There is the guilt of the knowledge that the disease is passed on through the parents. There is also the issue of carrier testing, when and is it should be carried out, and the implications of genetic conditions on the entire, extended families of people with Batten disease. This ties in with the arguments within the disability literature surrounding genetics and the development of therapies which aim for eradication of diseases of this type, along with the pressure on healthy carriers to decide whether or not to test foetuses or babies for the disease and what to do if the tests come out positive. Although not common to a wide range of chronic illnesses, these issues are common to chronic illnesses, both rare and common, that are genetic in nature.

Stigma

The third theme stemming from the research on chronic illness is that of stigma. Stigma has been identified as the potential spoiling of identity through the negative reactions of others to changes in normal bodily or behavioural experiences (Anderson and Bury 1998). Stigma is seen, in many cases, to stem from the application of a diagnostic label (Robinson 1988; Scambler 1984), and is at the centre of many of the interpretative accounts of living with a chronic

illness. Stigma has been referred to both in terms of the threat of stigmatisation ('felt' stigma) and actual episodes of discrimination ('enacted' stigma) (Scambler 1984). There has been no research looking specifically at the issue of stigma in this disease, although a number of the parents in the study carried out by Scambler (1999) identified stigmatising behaviour from family members when it was discovered that the condition was of genetic origin. One mother, for example, talked of trying to suggest to family members that they were tested to see whether or not they were carriers and being ignored. In addition, other parents reacted differently towards both the parents and the affected child when the terminal nature of the condition was more widely known. This reaction, along with that of the extended family, often made the parents and siblings feel more isolated and stigmatised. In terms of diagnosis, the interview data suggests that it is not the label of juvenile Batten disease which causes the distress of the families so much as the reality of the condition and its long-term prognosis.

Biographical Disruption and the Reconstitution of Self

Research shows that the diagnosis of chronic illness has a huge impact on the self-image of the diagnosed person, as well as on their public image, and causes them to rethink their past, present and future. Meanings change across time (Pinder 1998), and there is a need for negotiation and renegotiation (Locker 1983; Anderson and Bury 1998). The extreme biographical changes that occur from the onset of the symptoms of juvenile Batten disease, and the implications of this for the reconstitution of self are looked at in detail in a paper written by

Schroijen (1993), an educational psychologist working at the Bartimeushage centre in the Netherlands:

> "The children experience radical changes already in this period: psychological and medical tests; becoming blind; learning problems in the normal school; becoming an exceptional child; transition into a special school and maybe into an institution for the visually handicapped, where again they have to face learning problems; the uncertainty and growing realisation that they cannot do as easily what was possible before and that peers can do better; being captured by the first epileptic seizure." (p4)

These are only the changes that have to be faced in the first stage of the disease. These are followed by increasingly severe mental and physical deterioration, the loss of verbal communication and the decline into immobility. Shroijen suggests that the common reactions to these changes are denial, resistance, feelings of isolation and increasing dependence, combined with an awareness of age and self that continue as faculties deteriorate.

There is also the issue of maintaining an unchanging sense of self identity despite constantly readjusting to new realities, a concept described in the chronic illness literature as 'biographical disruption' (Bury 1998). This raises questions about the possibility of maintaining an unchanging sense of self when faced with both physical and severe cognitive degeneration. The interview data (presented in chapter five) however, suggests that both parents and professionals

strongly believe that the children are able to maintain an inherent sense of self-identity. And that the personality of the young people endures even as their physical and cognitive skills degenerate. Parents talked of their children refusing to 'give in' to the disease. They also spoke of a constant revisiting of favourite films and music, throughout the course of the disease. The degeneration of short term memory and retention of long term memory may, however, mean that the young people are simply holding on to what they can remember rather than expressing a sense of self and an active choice about who they are and who they want to be.

The diagnosis also has an extreme impact on the biography and identity of the family as a whole. Parents are required to cope with the loss of their 'normal' dreams and hopes for their child and themselves (Bowman, 1998). Losses include:

> "The dream for the 'normal' child...the dream about how a home 'should be'...the dream of time for self...the dream of family privacy...the dream that the couples relationship could withstand any stress or challenge...the dream of family...the dream of giving birth again." (p18)

In addition to this they have to come to terms with being the parents of a 'disabled child' and entering a world of specialists, hospital visits, special schools, statementing, social services, health services and residential care, all of which is overshadowed by the knowledge that their child's illness is terminal.

As previously outlined (in chapter five) two mechanisms are suggested by way of assisting young adults with juvenile Batten disease to maintain their self-identity, firstly through the shaping of present identity on past experiences, and secondly through the necessity of continuity of carers. It is at this point that the experiences of people with juvenile Batten disease again depart from those of the majority of people with more common chronic illnesses. The families and carers of the young people see the need to maintain the sense of self-identity of the affected young person for them as they are deemed unable to do so themselves. It is interesting to note that the young people are not deemed able to maintain their sense of self-identity in their own right and are seen to need the assistance of both family members and carers. Again this points to the thesis that the families are experiencing the condition along with the child, to the extent that they are actually instrumental in maintaining the 'self' identity of the child. It could also be argued that this is not infact a self-identity, but an identity imposed on the children by the people around them because of the children's inability to express their identity themselves. The loss of short-term memory also means that. Whilst negotiation takes place at symptom onset, and renegotiation occurs while the young person is deemed able to readjust, a point is reached where renegotiation is no longer deemed possible and repetition is used to maintain the former identity of the young person rather than seeking to renegotiate a new identity.

Managing the Medical Regimen

As illustrated in the literature review (chapter two) the issues around the management of the, often complex, medical regimen surrounding living on a daily basis with a chronic illness covers three main areas: balancing the costs and benefits of treatments; the loss of the time needed for medical treatment; and the need for negotiation between doctors and patients. All of these issues are relevant to the lives of families living with juvenile Batten disease. The need to balance the costs and benefits of treatment were highlighted by Ruth Pinder in her work on Parkinson's Disease (1998) where she explores the dichotomy of using drugs to combat the symptoms of the disease and the subsequent symptoms caused by the drugs themselves. Similar problems are found in juvenile Batten disease with the need to experiment with different drugs to control symptoms such as the, often acute, epileptic episodes, and traumatic hallucinations. Aside from the use of recognised drugs to control the main symptoms of the disease, the families also have to deal with issues around whether or not to try different food supplements and experimental drugs and which drug combinations work best. A whole range of treatments have been tried by parents and researchers seeking to understand and control the disease and, alongside the need to manage the standard drug regimen, families also have to decide which, if any, other experimental treatments they with to try. The Batten Disease Family Association is the formal forum through which parents talk to each other and recommend different treatments, but parents also talk informally about their experiences.

The problem of time management and loss of time to medical treatment, particularly highlighted by Jobling (1998) in his work on the, often intensive, treatment regimes followed by people with acute psoriasis, and by Locker (1983) who explored the time lost to other, more enjoyable pastimes, is also one experienced by families with juvenile Batten disease. The need for parents to manage medical regimes spans the management of epilepsy and hallucinations, feeding through a naso-gastric tube or gastrostomy, basic physiotherapy exercises and a whole range of personal care needs. Parents receive little or no training and are expected to manage whatever happens (Scambler,1999). Again, the accounts of parents trying to cope with the disease, explored in detail in chapter five, are around the accounts of mothers trying to cope with the medical needs of their children within the family home:

> They were giving me medicine and I was supposed to give him 11 mils of this medicine four times a day, and I've only got a 5 mil spoon. How do I give him 11? You know, so, and I could never give it to him at, say like 10 o'clock, 1 o'clock, 4 o'clock, cause he was never here, so it was just get it when you can. So in the end I used to give him four doses of 10 mils and then when he's go to bed I'd give him 15.

> I can only do so much, like physiotherapy, you know, I'm limited with the room...Sometimes he won't eat in the morning so I put him on the pump...I have to take the tube off and syringe it through and clean it, you know...I do everything, all the nursing care...I just

> picked it up as I went along. It's like when [he] started having fits, we went to the doctor and he was in hospital. I just get on and do it. It's like when he had his gastrostomy done, I was shown when he had it in the hospital, this is what you do...I was just shown once, you know, and I learnt it there.

Clearly these two young people are in the later stages of the disease process and have medical regimen which require considerable management. In both of these cases the mothers are trying to cope with little equipment and no proper training. These examples do highlight, however, the huge amount of time and effort needed to manage the condition, and also the considerable time lost to other, more pleasureable, family and individual activities.

The final theme to emerge from the literature was that of the need for negotiation and open communication between doctors and patients. Both Jobling (1998) and Scambler (198?) referring to Psoriasis and Epilepsy respectively, talk of the need for doctors to give adequate time to patients, to answer their questions honestly and to recognise the in-depth knowledge that their patients are uniquely in a position to collect. This is no less the case for parents of children with juvenile Batten disease. The severity of the disease means that parents will have a list of questions that they will want answers to, require the time and space to formulate the questions that they want answered, and need honest answers from their doctors. In addition, the rarity of the condition means that parents are likely to know as much, if not more than the majority of the medical professionals that they come into contact with. This

needs to be recognised if the medical profession is to make use, as it should, of the wealth of specialist knowledge that many of the parents have accumulated over the course of the disease. Information is collected by parents from a range of sources including medical libraries, the medical profession, charitable bodies, the BDFA, other parents and, more recently the internet. Consequently many of the parents are fully aware of the implications of the disease but also very knowledgeable about the latest treatments and trials being carried out. This makes them very useful resources for doctors if they are allowed to share their knowledge.

The main difference between the experiences of families with juvenile Batten disease and people with other more common chronic illnesses, aside from the severity of the symptoms of juvenile Batten disease, lies in the role of bio-medicine. The review of the literature suggests that the sociology of chronic illness developed almost as a challenge to the power wielded by a bio-medical profession which can, in reality, do very little for the majority of people living with chronic illnesses for which, at the present time, there are no cures. With juvenile Batten disease, bio-medicine is not just seen as the way of managing symptoms but is also giving hope to the families of the children. Advances in genetic research and mouse modelling are the only hope for these families and so many of the parents are happy, or even fighting, for the chance for their children to be involved in trials.

Information, Awareness and Sharing

The final theme identified is that of information, awareness and sharing. Within this theme there are three main issues: the quality of verbal and written information provided by doctors and other medical professionals; management of consultations and the power relations between the doctor and their patients; and the need to get information from alternative sources. All of these issues are as relevant to families with juvenile Batten disease as to people with more common chronic illnesses, and in some cases the lack of information is more severe because the disease is so rare. The relevance of each of these issues can be illustrated by taking each in turn and looking both at their impact in the wider literature, and, more specifically, at the role that each plays in shaping the experiences of families with juvenile Batten disease.

A significant number of the families interviewed in this study expressed dissatisfaction not only with the quality of the verbal and written information provided by doctors, but also with the accessibility of the information and the ways in which it was delivered. As highlighted in chapter four, the vast majority of the families were not happy with the amount of information made available to them, either at the time of diagnosis or later on. Even where information was made available it was often not in an ideal format and contained medical jargon unintelligible to parents who were not themselves in the medical field. A number of parents also talked about the starkness of information written in black and white and the lack of humanity in trying to come to terms with information of this kind with no explanation or opportunity for questions. Again this ties in with the wider literature not just on the

importance of providing information, but on how such information is communicated. Macdonald (1998) focused on the inadequate information received by people with rectal cancer and the crucial role that information played in reducing the impact of felt stigma. Anderson (1998) also echoed the issue raised in the study around inaccessible, jargonised information, suggesting that the information provided by doctors and medical professionals is focused primarily on physical impairment and rarely looks beyond that at the implications of the disease for people living with it on a daily basis. This leads directly to the need to get information from alternative sources.

As suggested in the previous chapter, the growth of self-help groups, covering the broad range of chronic illnesses and many other medical conditions, can be seen as a direct response to the inadequacy of information provided by the medical profession, reflecting the need for more, accessible, information. Whilst the majority of the families in this study did feel that they had enough information on the disease, most had had to compile the information for themselves. Parents attended conferences, accessed information in medical libraries and searched the internet to find what they needed. The main source of information, however, was other families, accessed both through the Batten Disease Family Association, through other charities and self-help groups both in the UK and abroad, and through common ground such as special schools. The self-help culture pervading the literature on chronic illness also plays a major role in the experiences of families with juvenile Batten disease.

There is clearly a lack of information on juvenile Batten disease for parents or professionals, due both to the nature and rarity of the disease. There is a minimal amount of medical information and expertise available and virtually no information on practical day to day issues of caring for a child with the disease. The families interviewed in the study by Scambler (1999) highlighted the need for basic information for medical professionals, and the struggle to get their acquired expertise acknowledged. They also stressed the need for more training and awareness on the part of doctors when giving the diagnosis in the first instance. This being said however, juvenile Batten disease is clearly not unique, and the lack of adequate, accessible information is also felt across many of the more common chronic illnesses. Awareness is growing, but at the present time very little information exists.

The final point to make when looking at the impact of information, awareness and sharing concerns the relationship between the patients and the medical profession in relation to the accessibility and reciprocal nature of information exchange. The chronic illness literature suggests, as argued in chapter two, that the traditional doctor patient relationship characterised by the all-knowing dominant doctor and the unknowledgable, subservient patient is being challenged. Research suggests that chronically ill patients in particular are dissatisfied with the relationships that they have with their doctors (Fitzpatrick 1990) and in particular with the amount of information received (Patrick et al 1983) and the way in which it is communicated (Harding and Modell 1985). In a study on epilepsy, Schneider & Conrad (1983) talk about the need for doctors and patients to develop a reciprocal relationship as co-participants in

care where each shares information and learns from the other. This is a view echoed by the families with juvenile Batten disease, with parents frequently commenting on their status as 'experts' on the disease and the fact that this status and the information that they have collected is rarely acknowledged in the course of formal doctor patient interaction. Conversely, however, a number of families talked of having built up close and reciprocal relationships with family GPs who had developed an expertise in the disease, often over the course of many years of contact with the child and their family, and who were open to and aware of the expertise gained by the parents.

It is clear from the above that there are many ways in which the experiences of people affected by juvenile Batten's disease reflect the experiences of people with other chronic illnesses. However, as illustrated in chapters four and five, there are a number of ways in which it differs. There is no expectation within juvenile Batten's disease that a 'normal' life is possible, there is no direct information sharing relationship between the chronically ill person and the medical professionals caring for them, most information on the disease and how it effects people on a day to day basis comes through indirect sources (parents, carers, teachers) as interviewing the young people themselves is problematic and, although often viewed as inadequate, there is the acknowledgement that bio-medicine is the only hope for any kind of future life for the chronically ill children. The relationship between chronic illness, juvenile Batten disease and bio-medicine is looked at in more detail in the next section.

Juvenile Batten Disease and the Challenge to Bio-medicine

An analysis of the literature around the sociology of chronic illness, as previously suggested, clearly illustrates the widespread negative view of biomedicine, not in terms of the interventions that it seeks to make so much as in those that it does not. There are three key areas in the literature around chronic illness and bio-medicine which can be differentially addressed through the looking at the experiences of families with juvenile Batten disease.

1. The sociology of chronic illness challenges the assumption that illness is bad and deviant and that people should want and are able to get better.
2. The Medical Professional is seen as inadequate because it focuses almost exclusively on bio-medical interventions and forgoes the other roles that it should perform.
3. The most accurate information about living with chronic illnesses is only obtainable through direct interviews with chronically ill people themselves.

Taking each in turn the areas of divergence and convergence can be illustrated.

The sociology of chronic illness, as argued in chapter two, developed through the realisation that the bio-medical model of health does not adequately account for the significant effect that chronic illness has on the lives of the sufferers and their families. Sickness was seen as an abnormal, transient state through which people should aim to pass as quickly as possible (Parsons 1951). This view caused problems across the gamut of chronic illness where, for the vast

majority of people, the sick, or impaired, role (Gordon 1966) is a permanent condition which cannot be transcended through the efforts of the individual and which does not necessarily bring with it the negative, stigmatising state identified by Parsons (Kassebaum & Bauman 1965, Sieglar & Osmond 1974, Barnes et al 1999). Families with Batten disease experience the same permanency of the sick-role status as people with diabetes, multiple sclerosis and any number of the, as yet, incurable chronic conditions. Once the diagnosis is given it is permanent and the status of the effected child, and, to a lesser extent, their family, is altered for the rest of their lives.

The first area of divergence in experience and views surrounds the perceived role of the medical profession and the significance placed on the development of biomedical interventions. The general literature places an emphasis on the non medical roles performed by the medical profession. In particular, attention is paid to the role of the doctor in sanctioning a range of services and benefits available to people living with debilitating illness over extended periods of time (Waitzkin 1989) and the role of chronically ill patients in challenging the, often unquestioned, authority of the doctors in these cases (Silverman 1987). Further to this, the lack of practical information and support provided by doctors, often despite their role as gatekeeper to a range of support and social services, is also highlighted (Jobling 1998, Cooper & Huitson 1986, Ziegler 1981, Anderson 1998) The families in this study also focused on the role of doctors as gatekeepers to other services and the lack of information available on non-medical aspects of care, support and the services available to support families living with conditions such as Batten disease. Many of the parents were not

even aware of the types of financial support available to them and the means of accessing them. The difference in experience, however, emergences when the primary role of bio-medicine is explored. All of the families in this study were, at the very least, aware of the genetic and biomedical research being undertaken in the area of Batten disease both in the UK and globally, and many were actively involved in one or more projects. This was seen by many as the primary role of biomedicine, and the only hope for any kind of future for children living with juvenile Batten disease. Thus, bio-medicine was primarily seen in a positive light and close relationships were developed between the researchers and practitioners and the parents, in contrast to the experiences of many of the people with more common chronic illnesses. That the relationship between the bio-medical/genetic researcher teams and the parents was seen, by both parties, as a positive development could be seen as due to the nature of the disease itself, but also, partly, as due to the rarity of the disease and the approachability of the team of researchers involved in this field.

The final area in which the common, primarily negative, approach to bio-medicine is challenged relates to data collection and the use of expert-led and user-led information. This is an issue explored in some detail in chapter four where the particular problems around researching juvenile batten disease are explored. It is widely felt that the only way in which the true extent of the impact of living with a chronic illness can be understood is through words and subjective experiences of the people living with the conditions. With juvenile Batten disease, however, as previously argued, this is not possible, both due to the impact of the symptom combinations which severely effects both

communication skills and understanding, and because of the lack of direct knowledge about the condition and its implications amongst those suffering from it. This means that expert-led professional accounts of the disease necessarily play a more significant role in our understanding of what the conditions means and how it effects the young people living with it. Whilst the accounts of families play a pivotal role in our understanding, the accounts of professionals cannot be discounted and thus, again, bio-medicine plays a more central role than it does in many other, less extreme conditions. Again these arguments can be extended across the experiences of children and young people with a variety of metabolic, genetic conditions which may also be categorised as chronic, are becoming increasingly common due to improved longevity, and also result in the same types of multiple, profound disabilities.

Juvenile Batten Disease and the Social Construction of Chronic Illness

The final area to be explored when examining the possible impact that the study of a condition such as juvenile Batten disease might have lies in the arguments around the social construction of chronic illness, and again hinge on the perceived importance of the bio-medical. The first point to make here is that the point of diagnosis, whilst both memorable and traumatic, is often of less importance in terms of labelling for people with juvenile Batten disease than it is for people with other less severe conditions. Within the chronic illness literature diagnosis is seen as a momentous occasion resulting in the labelling

of an individual as chronically ill and the possible change in their master status identity. With juvenile Batten disease the diagnosis, while clearly momentous in itself, and often long awaited, may come second to the severity of the symptoms already being experienced. By the time the majority of the children receive their diagnostic label they have already developed a number of symptoms and had to make the transition from a 'normal' to a visually impaired child, from there to a child with special educational needs and from there to a child in a special school. A number of labels have already been placed on the child, and, although clearly the most severe in terms of implications, the diagnosis is one label in a line of labels, all hinging on the developing symptoms faced by the child. There is no doubting, however, that each of the new labels placed on the child, and therefore, by association, on the family, is both traumatic in itself and has social implications in terms of the way in which both the child and the family are viewed by wider society. Whilst the shame and embarrassment associated with the labels (Nettleton 1995) are clear in terms of the stigma felt and faced by the family, the major impact of the disease is still manifested in terms of the bio-medical symptoms that have to be coped with by the child and their family on a daily basis. Thus, to suggest, as Barnes (1999) does in his review of the literature, that the impairment becomes secondary to the effect it has on discourse, is to minimise and trivialise the truly devastating effect that the symptoms of the disease have above and beyond the effects that discourse and language have.

Summary

This chapter explores the literature around the sociology of chronic illness in so far as it reflects the experiences of families living with juvenile Batten disease. The nature of juvenile Batten disease as a chronic illness is explored in terms of the degree of congruence between the experiences of the families in this study and those reflected in the wider literature. For each of the six key themes the areas of convergence and divergence are identified and discussed. Finally the role and perceived importance of bio-medicine are explored both directly, in terms of an exploration of the literature as a challenge to bio-medicine, and indirectly in relation to arguments around the social construction of chronic illness. This chapter makes the case that, whilst juvenile Batten disease clearly fits the criteria of being a chronic illness and shares many of the problems more widely associated with this group of conditions, there is a key area of divergence around the importance placed on bio-medicine both in terms of the daily lives of the families living with the condition, and in terms of their wider understanding of what the condition means and what its impact on them will be. The main arguments are represented in table 8, where the views from the literature on the sociology of chronic illness are added to those of the families and those suggested through bio-medical facts of juvenile Batten disease itself. In the next chapter the experiences of families living with juvenile Batten disease are explored in relation to the wider sociological literature on disability, and, again, the role of bio-medicine is discussed and challenged.

Table Eight

Biomedical Fact Of Juvenile Batten Disease	Issue Raised Through Experiences of Families	Issue As Seen Through The Chronic Illness Literature
Genetic Disease	Gene Replacement Therapy, The New Genetics, Impact on the Family	Genetic Nature Important in terms of Impact on Family, not In Itself
Very Rare Condition	Very Limited Literature, Small Sample Population, Methodological Difficulties	Not Covered in Literature, Vast Majority of Chronic Conditions more Common
Diagnosis is difficult	Rarity, Diagnostic Techniques Available,	Diagnosis often Difficult and Pivotal
Degenerative Disease	Constantly Changing Reality over a Relatively Short Period of Time	May be Constantly Changing Reality Across Time
Profound Multiple Disabilities	Severe Disability, Affecting Many Aspects of the Individual	Disability, Often Becoming More Severe, Affecting Many Aspects of the Individual
Severe Cognitive Degeneration	Gradual Loss of Skills, Focus on Maintenance not Learning New Skills	Cognitive Degeneration May or May Not Be Present
Short Term Memory and Communication Skill Losses	Loss of Means of Maintaining Sense of Self	Memory and Communication Skills Losses May or May Not Be Present
Bio-medical Care is Essential	Bio-medicine Gives Only Hope for Present and Future	Bio-medicine Often Of Little Relevance to Daily Life
Specialist Care Needed	Normalisation is not Possible	The Aim is to Live a Normal Life With the Condition
Family Role is Crucial	Carriers, Carers, Proxy Information, Live with the Disease, Continuity	Carers, Family also Affected but Sufferers Central

8 Juvenile Batten Disease and Changing Experiences and Understandings of Disability

In the previous chapter the experiences of families living with juvenile Batten disease were compared with those of people with a whole range of more common chronic illness through examination of the key themes emergent through the literature on the sociology of chronic illness. This chapter, similarly, explores the experiences of families with juvenile Batten disease in relation to the wider literature on disability and the changing understanding of what disability is and how and why it effects people living with a diverse range of physical and cognitive impairments. The debates around definitions and meaning; the development of a disability politics and debates around genetics and disability are explored with reference to the experiences of families living with juvenile Batten disease. These debates are then used to answer the research questions, raised in the introduction, and repeated below:

- To what extent does disability theory reflect issues pertinent to disability caused by chronic illness?
- To what extent does disability theory take into account the particular problems encountered by people with rare conditions

- To what extent does disability theory take into account the particular problems encountered by people with rare conditions and what explanations/solutions are provided for the particular issues these conditions raise?
- What account does current disability theory make of the issues faced by people with profound multiple disabilities and those assisting them with their care?

This chapter starts with a look at the debates around the social construction of disability and the role of impairment before looking at the growth of disability politics and the mirrored development of the Batten Disease Family Association. The debates around genetics and disability are explored with direct reference to the implications of genetic research and development for families living with juvenile Batten disease. The final section of the chapter then takes the preceding debates and relates them directly to the research questions above with the aim of summarizing the ways in which the experiences of families with juvenile Batten disease reflect and challenge the key developments in disability theory since the 1960's.

The Social Construction of Disability and the Role of Impairment

The central debate concerning the definition of disability, as catalogued in chapter two, has been ongoing since the late 1970's with the major arguments surrounding the medical and social models of disability and their

corresponding advantages and disadvantages as ways of understanding exactly what disability is and what it means to people living with disabilities. The definitional frameworks and their critiques are complex, hinging on the role of bio-medicine in defining and 'treating' disability and the role of society in shaping the experience of disability. It is this interface between the bio-medical and the social that is at the heart of the most recent debates around the introduction of a sociology of impairment. Whilst the impact of society and the environment on disability are acknowledged in the latest version of the International Classification of Impairment, Disability and Handicap (ICIDH2) advocates of the social model say that the latest changes do not go far enough and that disability has nothing to do with impairment and everything to do with the social environment and the ways in which it discriminates against non able-bodied people.

The argument for a sociology of impairment (forwarded by Oliver 1996) stems from the separation of disability and impairment into two quite distinct realms with disability falling firmly under the influence of social constructionism and discourse, and impairment being co-opted into the bio-medical sphere of influence. The three discursive mechanisms utilized for the separation of the two terms and the removal of impairment are itemised by Corker and French (1999):

- By 'the language and politics of exclusion' (Riggins 1997) or omission, which in linguistics is referred to as elision. Here, whereas 'impairment' is only an attribute or identity, disability is a

framework of differential analysis and a primary way of signifying relationships of power. Thus impairment becomes a fixed surface onto which disability is projected by 'culture'.

- Because impairment is also a referent for the 'individual', 'medical' and 'administrative' models of disability, this commonality must be denied. This strategy is implicit in the suggestion that we must not talk about impairment because this 'allows those who wish to see disability as personal, pathological and impairment specific an opportunity to use a misappropriation of the social model of disability as justification for preventing or blocking disability equality' (Darke 1998: 224).
- This is achieved through the production of a division of meaning (semantic splitting) of 'impairment' and the redistribution of its constituent parts in such a way that the boundaries between disability and impairment are blurred. For example, *disability* is more accurately described by terms such as wheelchair user, Braille reader and hearing aid user, and impairment by terms such as mobility impaired, visually impaired and hearing impaired. However, the latter terms are frequently used as descriptors for the former, thus conflating disability and impairment. (Corker and French 1999: 3)

They go on to suggest however, that, whilst Oliver's arguments about definition as more than semantics (1996) are valid, there is a danger that by discarding the whole of the disabled/impaired body to the realms of bio-

medicine, the body is entirely medicalised whilst disability is left in no-man's land. These arguments are echoed by Williams who suggests that by removing the body from the debate around disablement the social model of disability is moving in the opposite direction to the sociology of the body which proclaims to look at the same issues (Williams 1990), and is gifting the body to bio-medicine unchallenged.

Corker and French (1999) are in broad agreement with Hughes and Patterson (1997) who go further to suggest that if you follow these arguments to their logical conclusion:

> The relationship of disabled people to their bodies is mediated by medicine and therapy, and has nothing to do with policy and politics. This dualistic approach produces a theoretical rigidity which involves the medicalisation of disabled peoples' bodies and the politicization of the social lives. There is a theoretical closure around the relationship between sociology and the body which makes a sociology of impairment unthinkable." (p331)

From this point a new way of incorporating the body into discourses around disability is needed if the impact of the impaired body is to be incorporated into our understanding of disability. The boundaries between disability and the body need to be explored, and a common, middle ground sought. In the words of Corker and French:

> "...the presupposition that the boundary between disability and impairment is solid does not allow us to explore adequately our experience of disability oppression because this experience is 'in between' – discursively produced at the interface of society and the individual..." (1999: 4)

They go further to suggest that one might usefully draw on work around the sociology of the body and Shildrick's (1997) concept of 'leaky bodies and boundaries' to draw the two sides together and explore the, less than distinct, boundary between them.

It is this, seemingly artificial, separation of the impairment and the disability, the removal of the body from this branch of disability theory, which is most relevant to, and detached from the experiences of people living with juvenile Batten disease. Here too it could be suggested that the boundaries between the impaired body and experience of disability are less that rigid. If you take, as an example, the disease trajectory followed by a child with juvenile Batten disease, the links between, and impossibility of separating, the physical from the social become apparent. The permeable boundary between impairment and disability can be illustrated through the disease trajectory incorporating the various stages of physiological degeneration, the corresponding care requirements and the social impact of each change on affected child. Harry Schriojen, an educational psychologist working at Bartimeushage in Holland (The first centre of excellence in the care of children and young adults with all

forms of Batten disease) outlines the combined effects of biomedical, social and environmental effects of juvenile Batten disease on child with the disease:

1. First there is the stage of growing up normally, mostly without special difficulties for the parents to be worried about. From this normal development children already fairly know what goes on in the world.
2. Secondly there is the stage of the first symptoms of the disease, from a mean age of about seven, in which the visual impairment and learning difficulties manifest themselves. The children experience radical changes already in this period: psychological and medical tests; becoming blind; learning problems in the normal school; becoming an exceptional child; transition into a special school and maybe into an institution for the visually handicapped, where again they have to face learning problems; the uncertainty and growing realisation that they cannot do as easily what was possible before and that peers can do better; being captured by the first epileptic seizure.
3. Then we face the stage in which the children experience becoming seriously handicapped in several fields. There is a growing and hard confrontation with their mental and physical deterioration, they show denial and resistance to this experience. Verbal communication slowly diminishes by declining articulation and difficulties with respect to the input, processing and output from information by wordfinding and aphaisic problems, along with

experiences of not being properly understood. Acceptance of help concerning their daily-living-skills is something they have to learn gradually. Their motor functions are limited extensively, so that the use of an adaptable wheelchair has to be introduced. They will become incontinent. One sees them struggling heavily with all these disturbing experiences, each according to his own character. There is a considerable risk of gradually feeling left more and more alone.

4. In the end immobility and slowly increased physical vulnerability require a more intensive and good nursing treatment. There is a great danger of apathy and inactivity: on the one side by their growing withdrawal into themselves, on the other side because their feelings of being understood depend more and more and totally on the capacities of their families and the nursing staff to subtly grasp their tokens and signals. Notwithstanding we feel it is important to establish and maintain a warm living atmosphere, where there have been preserved rests of memories and recognitions. (Schroijen, 1993: 3-4)

Schroijen looks at the development of the disease in terms of the changes being faced by the child. The disease trajectory can, similarly, be looked at in terms of the impact that it has on the family. In doing so the effects of the impairment can be separated from the disability in totality in the extent to which the parents do not directly experience any part of the impairment and

yet clearly have to cope with the 'disability' or the social and environmental impact of living with a child with juvenile Batten disease.

The initial symptom of visual impairment first manifests itself when the child is approximately 6 years old. This is followed rapidly by increased clumsiness and problems with schoolwork such as speech difficulties, a decreasing attention span and deteriorating cognitive skills. At this time the child usually makes the transition from 'normal' to 'special' educational needs and the possible move to a special school, and the parents make the transition from being the parents of a 'normal' child to being the parents of a child with 'special needs'. At some point before or during the time when the child moves to a special school the parents will received the diagnosis of the disease. Many of the parents then talk about starting a prolonged grieving process which takes them throughout the next two decades to the death of the child. It is the nature of the symptoms and the inevitable degeneration to death that causes this prolonged grieving process. It is also at this stage that the parents take on the identity of parents of children with a terminal condition. This also brings with it the loss of the dreams that the parents had of the lives that their children would follow and the loss of their 'normal' family life.

The initial symptoms are compounded by increasing visual problems and the degeneration of the short term memory, movements become slow, and there is the onset of generalised seizures, cerebellar ataxia and intention tremor. This is often followed by a transition to residential education with the children

boarding at the schools during term time. For the parents this is a time when they make the transition from full-time to part-time carer and brings with it the negotiations over the role of the parents and family in the care of children resident and being cared for outside of the family home. It is also a further challenge to their ideals about what a 'normal' family should do, where they should live and who should provide the care. In addition to the degenerations described above, there is the onset of dementia, the development of psychological and behavioural problems and recurrent, and frequently extremely upsetting, hallucinations. These symptoms may lead to the next major transition period in care, which is where the child, or young adult, is no longer able to remain in full-time education and must leave the special school. For some children this decision is made because of the increasing medical needs that they face, and for some it is simply because they have reached the age of 19 and are no longer able to stay in the school environment. At this point the families face the decision of whether or not the child should be cared for at home or within a residential care unit. This transition is looked at in detail in chapter five and involves debates over whether the parent(s) are willing, or able, to provide the full-time nursing care that their children will inevitably need, whether they are able to take on the medical tasks that will be required of them and whether the house that they live in is adaptable, in addition to worries over funding, finding appropriate residential care placements and so forth.

The final stage of the disease brings with it the need for 24 hour nursing care and, eventually, terminal care, as "the patient lies curled up in bed, blind and

speechless, with strong extensor plantar reflexes, occasionally adopting dystonic postures. Mercifully the illness ends in 10 to 15 years." (Adams et al 1997). All but one of the families felt unable to talk, or even think, about this point of the disease. It was discussed only in so far as one mother referred to the fact hat her son would remain in his present residential accommodation for the remainder of his life. It seems to be the case that this is something that is not considered if at all possible until absolutely necessary, although three of the parents of older children did refer to the fact that increased nursing care would be needed and that they did not want their children to be moved around if possible at this stage of the disease. It is at this stage that parents confront the terminal nature of the disease for the second time and enter the grieving process for the second time.

Whilst to conflate impairment and disability would be to loose the separate identity and impact of each, to separate the nature of the impairment from the nature of the disability in a case such as this would seem to trivialise the true impact of both. In addition, the fact that the impairments can have such a profound effect on the lives of the families (almost a secondary or 'courtesy' disability) clearly illustrates the links whilst dismissing the conflation.

In addition to the argument fielded above, a challenge to the social construction thesis can be formulated simply by examining the assertion that disability has nothing to do with the impairment but is caused by an environment which fails to cater for the needs of non-able-bodied people, being totally biased towards those without impairments. Taken to its logical

conclusion this line of argument would suggest that disability could be eradicated if an environment were designed commensurate with the needs of people with impairments. If, for example, an environment was designed with all furniture and equipment accessible, all floors level, single story buildings or lifts e.t.c. then there would be no disability attached to being unable to walk as this would not be a pre-requisite of functioning 'normally' within the social environment. There is, however, (at least) one major problem with this argument. It is simply not possible to design an environment which would cater to all of the physical and social needs of people with conditions such as juvenile Batten disease which result in profound multiple disabilities. Not only are the impairments themselves so extreme as to render the individual unable to function without (at its most extreme) continuous assistance, but the impairments themselves are degenerative and change over time, concomitantly changing the environmental needs of the young person. Even were the changes merely taken to refer to changes in social, as opposed to social and physical, environment, effecting the way in which we see people and the attributes that are valued socially, the dementia, hallucinations, loss of short term memory and cerebellar ataxia would still mean that the young person would not be able to function independently no matter how they are viewed. Thus environmental changes, however configured, would not eradicate the disabilities faced by these people, and many others with similar combinations of impairments.

Disability Politics and the Growth of the Batten Disease Family Association

Oliver (1990) Highlights four types of organization which have developed around issues of disability: 'Partnership/Patronage' groups; 'Economic/Parliamentarian' groups; 'Consumerist/Self-help' groups and 'Populist/Activist' groups. Detailed descriptions of the key aims of these groups can be found in chapter two, however the types of group most relevant to the emergence of the Batten Disease Family Association (BDFA) are the Consumerist/Self-help group and the Partnership/Patronage group. These groups are described, by Oliver, as follows. Consumerist/Self-help groups are:

> "Organisations of disabled people; self-help projects; sometimes campaigning groups, or working in collaboration with local or voluntary agencies – e.g. Spinal Injuries Association, Derbyshire Centre for Integrated Living, Berkeley Centre for Independent Living". (Barnes 1999, adapted from Oliver 1990: 117-18)

And Partnership/Patronage groups are:

> "Organisations for disabled people; charitable bodies; provision of services (often in conjunction with statutory agencies); consultative and advisory role for professional agencies – e.g. Royal Association for Disablement and Rehabilitation, Royal

National Institute for the Blind, SCOPE, Rehabilitation International." (Barnes 1999, adapted from Oliver 1990: 117-18)

Although the BDFA is not an organization *of* disabled people *per se*, as argued previously, the families of children with juvenile Batten disease are so involved in the disease process as to almost be experiencing the disease themselves, albeit indirectly via the social rather than physiological effects of the disease. Thus, the BDFA can be more accurately defined as an organization *of* disabled people than one *for* disabled people as the group provides support, advice and information for the family unit as a whole, including both the disabled and non-disabled family members. The BDFA also provides information for families, medical professionals, special schools and carers about the implications of Batten disease and the special needs that children and families living with this disease have.

Much of the political activity of disability activists is not directly relevant to the experiences of children, and more particularly young adults, with juvenile Batten disease – issues around access to voting rights and Independent Living for example. Campaigns around civil and social right (Barnes 1999), however, have played an extremely important role in ensuring that the quality of life of vulnerable children and young adults with diseases of this type is protected. Providing a good quality of life becomes more important than simply prolonging life because it is medically possible, and steps are constantly being taken to ensure that quality of life is available to even the most profoundly disabled people.

Juvenile Batten Disease and the New Genetics

The third debate of direct relevance to the experiences of families with juvenile Batten disease concerns the arguments for and against the 'new genetics', particularly around the extent to which it can be said to be eugenic – i.e. a direct attempt to improve the human species by controlled breeding for desired inherited characteristics. Some of the most recent debate in this area concerns the use of genetic intervention in human reproduction, predominantly by means of screening programmes for genetic conditions which cause disability. Clearly this is directly relevant to families with juvenile Batten disease which is a genetic condition for which prenatal testing is available at the current time. There are two opposing narratives at work around the issue of screening and gene therapy. The first sees the 'new genetics' as heralding a new era of reduced human suffering and the first step in eradicating a number of 'genetic' conditions which cause disability and death, whilst the second sees it as a covert form of eugenics and a way of eradicating the 'problem' of disabled people. The relevance of these debates to juvenile Batten disease can be seen by examining in more detail the two opposing narratives at work and their implications for the future of children and families with the disease.

The impact of being diagnosed with a genetic condition are wide ranging, affecting siblings and, potentially, members of the extended family, as well as the child first diagnosed with the condition. The empirical data collected in this study examined the extent to which the implications of the genetic nature

of the disease were explained at the point of diagnosis, whether the families were given advice about the availability of carrier testing for siblings and others, at the wider impact of the disease on siblings where one or more child is affected and at how parents decide whether or not to get subsequent children tested when they receive their first diagnosis, and at whether genetic counselling was offered to the families. The view of the 'new genetics' as a positive development was one shared by all of the families involved in this study to the extent that they were aware that gene therapy was the most likely source of a cure for the disease. There were, however, differences in the degree to which families were willing to get involved in research, and the amount of information that they had on the most current developments. The families with younger affected children were more, although not exclusively, likely to be actively involved in the research and conversant with the latest research developments.

The key issue, inherent in the second narrative around the 'new genetics' and eugenics, of whether or not to have children and potential children tested for the disease was one which concerned all of the parents with younger children or wishing to have more children, or with older children who were potential carriers of the disease. This links with arguments in the sociological literature around the issue of providing screening for conditions where no cure is available except termination (Shakespeare 1999; Bailey 1996). The debate is encapsulated in the dichotomy, as described by Lippman (1994) between the right of the mother (and father) to choose whether or not to have screening and continue with a pregnancy where the disease is present, and the extent to

which this choice is subverted by the desire to rid the world of disabled people. There were differences in the choices made by the parents in this study, opinion was split over whether testing was a desirable option with two of the parents with younger children choosing not to have their children tested and one choosing to go ahead with the tests, but none were in the position of having to decide whether or not to have prenatal testing. This dilemma was more common for families with children with infantile Batten disease where the children have a younger average age of symptom onset and diagnosis. At least two of the families in the Batten Disease Family Association, with late infantile Batten disease, had had to make this decision.

It would seem likely that, whatever decision around testing had to be faced, whether related to prenatal testing, sibling testing or carrier testing, all of the parents would have been horrified at the thought that their actions and choices could be interpreted as part of a covert operation to rid the world of disabled people. This is particularly likely to be so in light of the fact that juvenile Batten disease is so rare that testing is only carried out where one positive diagnosis has already been made in the family. Thus, all of the parents in the position to make choices over screening already had at least one, dearly beloved, severely disabled child. Parents and siblings talked of the desire not to see another child suffer the bewildering and profound symptoms of the disease whilst being extremely thankful to have had the opportunity to get to know their child or sibling. This would seem to suggest that, in the eyes of families with this particular genetic disease, the 'new genetics' is a potential saviour, and the idea of being accused of, or linked with, eugenics would be

both abhorrent and inaccurate. At the same time, there is little doubt that the nature of juvenile Batten disease is such that if given the choice to have a child with or without the condition the results would always be in favour of eradicating the disease.

Chronic Illness, Rare Conditions and Profound Multiple Disability and Their Place in Current Disability Theory

Having assessed the extent to which the debates currently raging within the sociology of disability are relevant to, and reflect the experiences of, families with juvenile Batten disease, the final section of this chapter seeks to utilise the arguments already explored, along with the wider literature, to answer the three research questions posed in the introduction to this chapter:

- To what extent does disability theory reflect issues pertinent to disability caused by chronic illness?
- To what extent does disability theory take into account the particular problems encountered by people with rare conditions and what explanations/solutions are provided for the particular issues these conditions raise?
- What account does current disability theory make of the issues faced by people with profound multiple disabilities and those assisting them with their care?

The first question relates to the extent to which sociological theory around disability reflects issues pertinent to disability caused by chronic illness. This is particularly relevant in light of the fact that current estimates suggest that approximately 10% of the world's population, more than half a billion people, are disabled. It is predicted that this number will rise dramatically in the next quarter of a century (International Disability Foundation 1998). In addition to this, the advances in the treatment of acute conditions, advances in life preserving treatments and an ageing population all mean that the number of people living with chronic illnesses is increasing. The relevance of increasing levels of chronic illness to the literature around disability lies in the fact that chronic illness is the most common cause of impairment in the developed world with arthritis being identified as the key cause of disability in the UK (Martin et al, 1988) and 20% of people in the UK reporting limiting long-standing illness in 1998-99 (ONS 2001). In addition, there are 1300 metabolic diseases of childhood, many of which have combinations of symptoms similar to those experienced by children and young adults with juvenile Batten disease and many of which are also experiencing the same extension of life expectancy as treatments and interventions improve. This means that, along with an increase in chronic conditions, there will be an increase in the number of rare chronic conditions of this kind and consequently, an increase in the number of people living. Although chronic illness is the most common cause of disability in the developed world, however, it is important to note that 80% of the people in the world with disabilities actually live in the developing world where poverty, accidents and war are far more prevalent causes of

disability than chronic illness, and people with rare, extreme chronic illnesses and profound, multiple disabilities are far less likely to live into adulthood.

Existing theory around disability is almost entirely biased towards the experiences of disabled people living in the developed world with work focusing on definitions, equality, political action and civil and human rights. In this context it is clear that impairments caused by chronic illnesses, whether common or rare, should be included in the theoretical developments that are taking place. What is equally clear is that this is not the case. If the example of the main body of work around definitions of disability and impairment is taken, it seems that neither the 'International Classification of Impairment, Disability and Handicap' (ICIDH) nor the 'International Classification of Functioning, Disability and Health' (known as ICIDH2) take into account the experiences of people with impairments or functions of the body system which fluctuate over time or degenerate. Similarly, the social model of disability, as explored earlier in the chapter, fails to acknowledge that there are people with impairments which fluctuate, degenerate or are simply too complex and profound to allow for the development of an environment which would eradicate the disability that these people face. In the argument above, the example of juvenile Batten disease was taken, however, the same argument could be made for people with more common chronic illnesses such as multiple sclerosis or arthritis whose needs can change on a daily or even hourly basis. This would suggest, therefore, that whilst chronic illness is only a key cause of concern for disability theorists working with the framework of the developed world, the fact that the majority of the theory currently being

developed is commensurate with the experiences of this minority means that an understanding of the impact of chronic illnesses should play a significant role in the development of theory around disability. In addition, it seems clear that the number of people living with profound, multiple disabilities will increase over the next few decades as medical interventions continue to develop and, thus, more attention will need to be paid to those people living with disabilities for whom the goal of a 'normal', agentic, political life is near, if not totally unreachable. A final point to make when taking the case of people with profound, multiple disabilities is that, in many cases, independent living and community living actually result in less, rather than more, independence and quality of life as the services needed on a daily basis are often inadequate or unavailable within the community and require the use of specialized services with expert staff.

Summary

In this chapter the key debates and developments in disability theory have been explored in relation to their relevance to the experiences of people with juvenile Batten disease, and, more widely, to the experiences of people with a whole range of rare and more common chronic illnesses and complex disabilities. The arguments postulated suggest not only that much of the debate around disability theory is not directly relevant to people with juvenile Batten disease, but that the literature also fails to address the issues faced by a whole range of people with disabilities which are caused by chronic illnesses or are faced in combination. The main arguments from this chapter are

represented in table nine where the most commonly expressed views within this literature are compared with those found in the literature on chronic illness, the experiences of families with juvenile Batten disease and the bio-medical facts at the heart of the disease. The final chapter assesses the arguments made throughout this thesis in terms of the research questions posed in the first chapter. Each question is answered with the aim of reaching a conclusion as to how much an understanding of the experiences of families living with juvenile Batten disease can tell us about existing problems with the sociological understanding of chronic illness and disability, and the extent to which the experiences of this group are representative of the experiences of chronically ill and disabled people in general.

Table Nine

Biomedical Fact Of Juvenile Batten Disease	Issue Raised Through Experiences of Families	Issue As Seen Through The Chronic Illness Literature	Issue As Seen Through The Disability Literature
Genetic Disease	Gene Replacement Therapy, The New Genetics, Impact on the Family	Genetic Nature Important in terms of Impact on Family, not In Itself	'New Genetics' is Either The Miracle Cure All or Eugenics in Disguise
Very Rare Condition	Very Limited Literature, Small Sample Population, Methodological Difficulties	Not Covered in Literature, Vast Majority of Chronic Conditions more Common	Not Covered in a Literature Which Focuses on 'Simple' Disabilities
Diagnosis is difficult	Rarity, Diagnostic Techniques Available,	Diagnosis often Difficult and Pivotal	Diagnosis is Not an Issue
Degenerative Disease	Constantly Changing Reality over a Relatively Short Period of Time	May be Constantly Changing Reality Across Time	Degenerative Impairments Barely Covered in Literature
Profound Multiple Disabilities	Severe Disability, Affecting Many Aspects of the Individual	Disability, Often Becoming More Severe, Affecting Many Aspects of the Individual	Profound, Multiple Disabilities Barely Covered in Literature
Severe Cognitive Degeneration	Gradual Loss of Skills, Focus on Maintenance not Learning New Skills	Cognitive Degeneration May or May Not Be Present	Cognitive Impairment only Covered In Relation to Learning Impairment
Short Term Memory and Communication Skill Losses	Loss of Means of Maintaining Sense of Self	Memory and Communication Skills Losses May or May Not Be Present	Memory and Communication Skills Losses Barely Covered in Literature
Bio-medical Care is Essential	Bio-medicine Gives Only Hope for Present and Future	Bio-medicine Often Of Little Relevance to Daily Life	Impairment Removed to Domain of Bio-Medicine, Not Relevant to Disability
Specialist Care Needed	Normalisation is not Possible	The Aim is to Live a Normal Life With the Condition	Any Care to be Controlled by Disabled Person in Community
Family Role is Crucial	Carriers, Carers, Proxy Information, Live with the Disease, Continuity	Carers, Family also Affected but Sufferers Central	Non-Disabled People Cannot Understand What it is Like to Have Live With A Disability

9 Is Juvenile Batten Disease a Challenge to the Existing Sociological Understanding of Chronic Illness and Disability?

This final chapter seeks to draw all of the strands of the arguments presented in the thesis together, with the ultimate aim of attempting to answer the main research question posed in the introduction to this thesis:

> *Is it necessary to develop an alternative approach to the theorising of chronic illness and disability to reflect the experiences of the many groups of people with rare chronic conditions who do not appear to be covered in the existing approach?*

In order to answer this question the arguments raised within the thesis are addressed. The issues covered are:

- Exploring the extremes of the chronic illness and disability literatures
- Rare conditions

- Rare conditions
- Embodied social actors
- The use of Critical Realism
- Developing a critique of the chronic illness and disability literatures
- A new theoretical paradigm

These issues illustrate the ways in which the experiences of families with juvenile Batten disease are relevant in a wider context. The issues are then readdressed in relation to the future of chronic illness and disability and the lessons that can be taken from this study.

It's clear from the analysis of the key themes and how they relate to the existing knowledge base within the bio-medical, sociology of chronic illness, and disability literatures, that there are a number of gaps in our understanding of what it is like to live with a chronic disabling condition such as juvenile Batten disease. The experiences of these families in particular are not currently incorporated in our wider understanding, a situation which this study hopes to go some way towards remedying. In addition there are some key themes and experiences which are either not represented in the literature in any form or are only touched upon – relating particularly to the extremes of conditions such as juvenile Batten disease and the place of rare conditions within our understanding. The aim of this study is to go some way towards filling this gap and suggesting possible areas for future research and theoretical development. The extent of the knowledge gap and possible future strategies can be illustrated by an analysis of the answers to the four research

questions introduced at the start of the thesis. Each of the questions has been addressed in the appropriate chapter, however, briefly revisiting each question and the problems raised in seeking to answer them illustrates the need to devote some attention to the final research question on the future of the sociology of chronic illness and disability. In the final two sections of this chapter we revisit the initial research questions before turning our attention to the larger question as to the future development of the sociologies of chronic illness and disability.

Issues Raised Through the Example of Juvenile Batten Disease

A summary of the convergence and divergence of experiences and opinions on the ten key issues as laid out in the introduction and explored at various points within the thesis is carried out below. For each issue a summary of the views and arguments from each sector of literature is provided.

The Genetic Nature of Juvenile Batten Disease

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Genetic Disease	Gene Replacement Therapy, The 'New Genetics', Impact on the Family	Genetic Nature Important in terms of Impact on Family, not In Itself	'New Genetics' is Either The Miracle Cure All or Eugenics in Disguise

Juvenile Batten disease is universally recognised as a genetic disease. In the bio-medical literature this is explored in terms of the autosomal recessive heredity and the fact that there is a 25% chance that each child born to carrier parents will develop the disease. The implications of this for the family are referred to in terms of the advisability of genetic counselling, the availability of both prenatal and carrier testing, and the scientific research aiming to identify the defective gene which causes the disease. The families interviewed through the course of this study then demonstrated this by talking about their hopes for current research into gene replacement therapy and the impact that receiving a diagnosis of a genetic disease has not just on the immediate family but on the whole extended family, raising issues about carrier testing, guilt and blame. Despite the importance of the genetic nature of the condition and the fact that a considerable number of, predominantly rare, chronic illnesses are caused by genetic defects, the genetic nature of some chronic illnesses is only referred to in passing in the literature in relation to the, admittedly significant, effect that this has on the wider family (as explored in detail in chapter two). Mention of the positive role of bio-medical research is negligible. The disability literature is split into two camps with one heralding the 'New Genetics' as the saviour of mankind (Conrad and Gabe 1999) and the other, majority camp, focusing on the 'New Genetics' as a form of eugenics and a covert way of ridding the world of disabled people (Shakespeare 1999). A significant part of the literature explores the developments around diagnosis and prenatal testing in relation to the desire to eradicate disabled people rather than as a step towards an ultimate cure for genetic disease. It seems clear from this analysis of the differing views that sociology has failed to explore

the impact of genetic disease on families in terms of the experiences and views of the families themselves.

Implications of a Rare Condition

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Very Rare Condition	Very Limited Literature, Small Sample Population, Methodological Difficulties	Not Covered in Literature, Vast Majority of Chronic Conditions more Common	Not Covered in a Literature Which Focuses on 'Simple' Disabilities

The medical literature on juvenile Batten disease estimates the prevalence of the disease as an average of 1.5 to 2 cases of NCL (Neuronal-Ceroid Lipofuscinosis or Batten Disease) per million children born in Europe. Figures based on diagnosis within the UK go on to suggest that there are approximately 10 to 15 new cases of NCL diagnosed each year, and thus there are between 100 and 150 children affected by one or other form of NCL in the UK at any one given time (Goebels et al 1999). The rarity of the disease is looked at in terms of the small number of people with expertise in the area and the lack of adequate specialized care placements for young adults with the disease. The rarity of the disease has a whole range of implications for families living with juvenile Batten disease, the key effects being the severe lack of information on the disease and particularly on practical issues, the lack of experienced carers and professionals and the need to continuously provide information for the medical professionals involved in caring for the children. In addition to this, in terms of researching the disease, the limited sample

population and the complex inter-relations between the families and professionals working in the area raise a whole range of methodological issues addressed both in chapter four and in the previous section. Neither the literature on chronic illness nor that on disability have a great deal to say about Very limited Not covered in

the particular issues facing people with rare conditions – although there is a body of work looking at the experiences of people with Cystic Fibrosis, which is a relatively rare condition. This is something that will increasingly need to be addressed as the number of children with rare conditions surviving into adulthood increases with improvements in medical interventions.

Experiences of Diagnosis

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Diagnosis is difficult	Rarity, Diagnostic Techniques Available,	Diagnosis often Difficult and Pivotal	Diagnosis is Not an Issue

The medical literature on juvenile Batten disease refers to diagnosis merely in terms of the various, both pre and post natal, diagnostic techniques available for clinicians seeking to make a diagnosis. They also refer to the fact that ophthalmologists are often the first to suspect the diagnosis and refer the child for further tests and that, due to the non-specific presenting symptoms, diagnosis is very difficult and relies on recognition of the signs. The experiences of the families reflect the views of the medical literature to the extent that the difficulty of reaching a diagnosis is recognised. Many of the families experienced delays in diagnosis and spoke of the, often traumatic,

array of tests that their children were subject to before a diagnosis was reached. They also spoke of receiving mis-diagnoses and a lack of information. The main theme to permeate the experiences of the families reflected the poor way in which many of the families were given the diagnosis. They spoke of a lack of information, doctors with poor communication skills and inadequate support either pre or post diagnosis. It was, however, noted that the situation was improving and that the experiences of families diagnosed more recently were often better than those of families diagnosed over the last two decades. The final point raised by the families in relation to the diagnosis concerned the implications of a genetic diagnosis on the wider family, something touched on in the previous section.

The impact of diagnosis was also reflected in the literature on chronic illness with diagnostic process often entailing a number of tests and an extended period of time, particularly in the case of diseases with a non specific selection of symptoms such as multiple sclerosis (Robinson 1988, 1998). The similarity with the experiences of families with juvenile Batten disease here is clear. However, the difference lies in the extent to which the diagnosis impacts on the identity of the sufferer. The literature focuses on diagnosis as a pivotal moment when the very identity of the individual changes and they go from being 'normal' to being a person with a chronic illness (Bury 1998, Robinson 1988, Scambler & Hopkins 1986, Schneider & Conrad 1983). This is not a view upheld by the families in this study where, although diagnosis is important and traumatic, it is not pivotal in the sense that all else revolves around it, because by the time the diagnosis is received the majority of the

families are already living with a whole range of physical and cognitive difficulties which balance the impact of the diagnosis and are more pivotal to their experiences. Diagnosis is not an issue in the disability literature as most of the disabilities referred to are either congenital or caused by accidents.

Degeneration

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Degenerative Disease	Constantly Changing Reality over a Relatively Short Period of Time	May be Constantly Changing Reality Across Time	Degenerative Impairments Barely Covered in Literature

Again the medical literature is straight-forward concerning the degenerative nature of juvenile Batten disease and cataloguing the faculties that will be lost and the approximate pattern in which the degeneration occurs. For the families the experience of degeneration is referred to more in terms of the uncertainty of never knowing quite what is going to happen next than in terms of physical losses, although major changes – for example from occasional to permanent wheelchair use – are noted. The families focus on the difficulty of adjusting to a constantly changing reality, and the practical and emotional difficulties of living with a disease which does not follow a specific pattern and worsens over the course of time, sometimes in gradual stages and sometimes by way of a sudden sharp decline. This is a theme that recurs time and again throughout the literature on chronic illness with uncertainty being one of the key themes to permeate the experiences of people living with a

whole range of chronic illnesses (Aggleton & Thomas 1988, Roth 1963, Anderson, 1998, Pinder 1990). Uncertainty relating to symptoms, trajectory and aetiology are noted and reflect the experiences of families living with juvenile Batten disease. The Disability literature, however, barely covers degeneration, being almost solely focused on disabilities which remain static across time.

Living With Profound Multiple Disabilities

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Profound Multiple Disabilities	Severe Disability, Effecting Many Aspects of the Individual	Disability, Often Becoming More Severe, Effecting Many Aspects of the Individual	Profound, Multiple Disabilities Barely Covered in Literature

The profound multiple disabilities referred to in the medical literature are catalogued according to the type of disability and the part or function of the body affected. Thus, visual impairment; motor degeneration – ranging from abnormal patterns of movement to wheelchair use to the loss of the swallowing reflex and incontinence; speech deterioration and the loss of control of facial expression; the onset of frequent and severe epileptic seizures; cognitive degeneration; short-term memory loss; dementia; and hallucinations are just some of the disabilities catalogued. Details on medical management are given in the form of symptom control and medical care needs. For the families the disabilities faced are no less real but are expressed in different forms. Parents talk about the activities that their children are no

longer able to participate in; the need to move to special schools and possibly even to permanent residential care placements; of having to fight for financial aid to adapt houses and the difficulty of seeing their children go through all of the, inevitable, degenerative stages of the disease. The human aspect of the multiple disabilities faced by people with a range of chronic illnesses are reflected in the literature where the effect of disability on many aspects of the life of the individual and their family are noted. This is particularly the case for people with illnesses such as multiple sclerosis (Robinson 1988) and Parkinson's disease (Pinder 1990) which have the same kind degeneration with a corresponding increase in the disability faced. Again, however, the disability literature barely covers the impact of living on a daily basis with profound multiple disabilities, focusing more on people who are ale to be fully agentic aside from having a disability (see chapter two for full argument).

Living With Cognitive Degeneration

Bio-Medical Opinion	**Family Experiences**	**Chronic Illness Lit**	**Disability Lit**
Severe Cognitive Degeneration	Gradual Loss of Skills, Focus on Maintenance not Learning New Skills	Cognitive Degeneration May or May Not Be Present	Cognitive Impairment only Covered In Relation to Learning Impairment

Problems with cognition are, again, catalogued in the medical literature. Cognitive problems are linked to irrationality, incoherence and the loss of awareness of reality. There may also be aphasic problems and a decreasing ability to communicate, with slower, unclear speech, stammering and a

reduced vocabulary – compounded by the deterioration of the speech motor system. Several years after the onset of cognitive problems dementia also begins to develop, resulting in a deterioration of, once 'normal', cognitive ability (unlike learning disabilities of difficulties where development does occur, albeit at a slower rate) and is gradual and irreversible. Dementia and cognitive degeneration are reported as leading to a deterioration in learning ability, social skills and emotional development. It is, however, noted that memories are still present although they are often vague, and children with juvenile Batten disease do have some awareness of the fact that they are growing in size and age despite the dementia. In addition, they are able to comprehend situations and sense moods but are unable to respond to them as their communication skills deteriorate.

For families there is the dual problem of coping with the gradual loss of skills and the fact that, as one parent notes, in the end it is like being stuck in a time warp with a child who has the mind and desires of a six year old forever, and the need to try and explain to the children why it is that they are not able to do the things that they were once able to do. This is particularly difficult where younger siblings are not affected and gradually overtake their older affected siblings both physically and mentally. Difficulties in explanation are exacerbated by the fact that none of the parents in the study had explained to their children what the consequences of their disease are and so each new development had to be interpreted separately. The chronic illness literature does not look specifically at the impact of cognitive degeneration although it is present in some of the conditions that have been studied, and particularly in

the work around dementia. Similarly with the disability literature there is little general work on cognitive degeneration although a considerable body of work exists, in parallel as opposed to integrated in the main body of work, looking specifically at the problems faced by people with learning disabilities.

Maintaining a Sense of Self

Bio-Medical Opinion	**Family Experiences**	**Chronic Illness Lit**	**Disability Lit**
Short Term Memory and Communication Skill Losses	Loss of Means of Maintaining Sense of Self	Memory and Communication Skills Losses May or May Not Be Present	Memory and Communication Skills Losses Barely Covered in Literature

Although the 'maintenance of a sense on self' is not referred to directly in the bio-medical literature on the symptoms and development of juvenile Batten disease, the means through which self identity may be lost are noted. Short term memory loss and the loss of verbal communication skills through a combination of a deterioration of the speech motor system, aphaisic problems and a decreased vocabulary all contribute to the loss of the means of maintaining and portraying a sense of self. These losses are referred to more directly by families in terms of the young person being unable to express their needs, likes and interests verbally, and having to rely increasingly on non-verbal signs, and past preferences. History becomes the present as the predominant links to the character of the young person in the present come

from a combination of knowledge of their past and best guesses. The mechanisms which precipitate the loss of self in juvenile Batten disease are common to other chronic illnesses. People affected by Motor Neurone Disease, for example, will gradually lose their speech motor system although the memory stays intact. Memory loss is also a key component of Alzheimers Disease which has been looked at in some detail. The specific impact of memory loss and speech loss are only part, however, of the literature on maintaining a sense of self which is a key theme in the sociology of chronic illness and is predominantly focused on the need to reinvent self-image, either once or continually, as the disease progresses. The need to 'cope' with a new 'spoiled' identity (Bury 1998) is a consistent theme throughout the literature on chronic illness, disability and juvenile Batten disease, although whether this change needs to come from within the individual or through a change in the attitude of the wider population, or, most likely, through a combination of the two, is up for debate. The difference apparent in the experiences of families with juvenile Batten disease is that this change in identity, identity recreation, is carried out by the family rather than the individual or wider society. It is the family who recreates the identity of the child through the use of diaries, and the maintenance of longer term memories where the short term memory no longer works. Therefore, although the specific medical problems noted in this disease are not necessarily covered in great detail, the overall effect of a loss of self and the need to reconfigure self-identity is explored in some detail in both literatures, although with different actors involved in the process.

The Role of Bio-Medicine

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Bio-medical Care is Essential	Bio-medicine Gives Only Hope for Present and Future	Bio-medicine Often Of Little Relevance to Daily Life	Impairment Removed to Domain of Bio-Medicine, Not Relevant to Disability

The bio-medical literature on juvenile Batten disease clearly states the need for specialist medical care throughout the course of the disease, increasing to 24 hour care in the latter stages. This care should be provided by trained, specialised personnel. Although the fact that there is no cure is acknowledged, the potential for a future cure is also reiterated and reinforced by the research currently underway both in the UK and abroad developing a mouse model of the disease and exploring the possibility of gene replacement therapy. For the parents, although there are many problems, particularly around the area of communication and information exchange, bio-medicine is largely seen as an ally – albeit an often ill-informed, non-communicative one. Research developments are heralded and most of the parents are extremely well informed on current and possible future developments in genetic and biochemical research in the area. Many of the families are also actively involved in one or more project, whether it involves giving blood, having tests, trying new drugs or using food supplements.

This positive view of bio-medicine is at odds with the current trends in both the chronic illness and disability literatures which see bio-medical intervention

as, at best, ineffective (Barnes et al 1999) and at worst a key cause of the disability faced by people with a range of impairments (Oliver 1990, Swain 1993). This is perhaps the most stark difference between the experiences or attitudes of families living with juvenile Batten disease and people living with a whole range of other more common chronic illnesses. The focus of the families in this study was two-fold. Maintaining a good quality of life was a central part of their goal, but they also remained focused on the only hope for the future in the form of the possibility of current research finding a cure for the disease. The literature on chronic illnesses is predominantly concerned with how people living with chronic illnesses cope on a day to day basis with the disease and bio-medicine is seen as something which, at best, is helpful, but at worst is ignorant of the holistic effects of the condition and unhelpful in dealing with these effects. One of the key current debates within the disability literature is looking at whether impairment has any place at all in discussions on disability (Oliver 1996, Corker & French 1999). And whether the impaired body should be left solely in the hands of bio-medical practitioners and excluded from discussion around rights and daily life with disability (Williams 1999). These arguments have been explored in detail in the previous two chapters. There is a clear difference between the views which predominate in the literature and those held by families living with this particular disease.

Specialist Care Needs

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Specialist Care Needed	Normalisation is not Possible	The Aim is to Live a Normal Life With the Condition	Any Care to be Controlled by Disabled Person in Community

Specialist care is needed throughout the cause of juvenile Batten disease and increasingly towards the latter stages of the disease. The bio-medical literature recommends seeing an ophthalmologist, paediatric neurologist, physiotherapist, occupational therapist, speech and language therapist, rehabilitation worker and counsellor at various points of the disease trajectory. The families also suggested that specialist care was needed although they suggested that it was often the amount of knowledge that an individual carer or therapist had about the disease and the amount of experience that they had of providing care for people with juvenile Batten disease that made the most difference. The majority of the families felt that the most appropriate care placement for their children, at least in the latter stages of the disease, was a residential placement in a home specifically catering for young adults with juvenile Batten disease and incorporating all of the facilities and expertise needed by this group of young people. At least two of the families, however, felt that the best place for their child to be living was the family home and that this could provide the most appropriate specialist care with the right support. All of the families agreed on the need for accessible specialist services such as physiotherapy and hydrotherapy, whether these were to be provided in day

centre settings or residential care settings. It was widely felt that residential care provided the most appropriate environment because it could be specifically adapted to the needs of people in wheelchairs or even staying in their beds but still being able to move around the home and take part in activities. (For an example of the development of a home of this kind see appendix one where a full outline of the development of Heather House and the services provided there is given).

The chronic illness literature focuses on the aim of living a 'normal' life with a chronic condition. This incorporates living in the community as an active part of the community. Again this is something that might be achieved in both a family home and a sensitively run small group residential home. The arguments around the provision of care within the disability literature focus on the amount of control that the disabled person has over the care that they receive (Case 2000; Oliver and Barnes 1998; Dale 1996). Again care within the community is advocated and care controlled, and preferably directly purchased by the disabled person themselves is seen to be the ideal. This fits in with the ideals of care suggested by the families, carers, teachers and doctors who took part in this study. Although care is often provided in residential units, the preference would be to have small group homes both located within and a part of the local community. In addition the ethos is to provide care appropriate to the needs and desires of each individual living within the home. Whether this is, in practice, the way that care is carried out, this would be the ideal for many of the parents and professionals that we talked to. However, the one caveat, from the viewpoint of the parents in the

study, would be that parents remained equal partners in the care and active decision makers in any and all of the major decisions made about the care of their child. The desires of the affected young person in this case are often represented by the parents and family members as the young person themselves is often unable to communicate what they like and want and so it falls to the family, who know them best, to fill in the gaps and make educated guesses. This ties in with the final theme which looks at the role of the family.

The Role of the Family

Bio-Medical Opinion	Family Experiences	Chronic Illness Lit	Disability Lit
Family Role is Crucial	Carriers, Carers, Proxy Information, Live with the Disease, Continuity	Carers, Family also affected but Sufferers Central	Non-Disabled People Cannot Understand What it is Like to Have Live With A Disability

The final, all pervasive, theme to emerge through both the bio-medical literature and the experiences of the families was the importance of the role played by the family. The literature stresses the role of the family as a partnership with the professional carers. In addition it mentions the importance of recognizing the impact of a genetic condition of this type on the extended family and raises the issue of genetic counseling and carrier testing for non affected family members. The evidence from the empirical data in this study suggests that the family of people with juvenile Batten disease are far more that simply partners in care. It was suggested, in chapter five, that the families could be seen as sufferers of the disease, not symptomatically, but

both directly as carriers of a genetic condition, and also indirectly through the impact of living in an 'abnormal' family. It was also suggested that many of the themes faced by the young people: uncertainty; the need to reconstruct self-identity and family identity as 'abnormal; the need to become involved in managing a medical regimen and to handle complex medical information are all faced by the family as well as by the individual. It could even be argued that, as the affected young people are not aware of the diagnosis that they have been given, the families suffer these things in proxy. The role of family and the impact of living with a person with a chronic illness are also key themes within the chronic illness literature with a large amount of work looking at the impact of chronic illness on the family as a whole, in terms of courtesy stigma, adaptations to the home, changing family roles and so forth (Goffman 1968, Robinson 1988, Anderson 1998). The disability literature, however, is largely working from a difference stance. It has been argued, predominantly by people from the social model standpoint, that the only people who can really understand what it is like to have a disability and to live in a non-disabled world as part of a minority group. Some theorists have gone so far as to suggest that only disabled people should be able to carry out research on disability because only they can truly understand what it is that they are looking at (Oliver 1990). Clearly this raises a number of problems for cases such as juvenile Batten disease where the affected children and young adults are not only unaware of the fact that they have the condition but would also be both physically and cognitively unable to carry out the research. This would mean that the only people qualified to carry out such research could not do so

and, therefore, that there would be no research on juvenile Batten disease or any of the many conditions with symptoms of this type or severity.

Having assessed the evidence within the thesis in relation to the key themes identified in chapter one it is useful to reiterate the answers to the research questions posed within the thesis as stepping stones in considering the ultimate aim of the thesis, an analysis of the impact of juvenile Batten disease on a sociological understanding of chronic illness and disability.

Answering the Research Questions

1. ***What is the understanding of chronic illness and disability within sociology?***

The current understanding of chronic illness and disability is explored in detail in chapter two where a thorough literature review is provided. The key to the current approach to the sociology of chronic illness is the inherent challenge to biomedicine. The underlying theme is the acknowledgement that bio-medicine can do very little for the majority of people with chronic illnesses living on a day to day basis with their condition as there is, most often, no cure. Similarly, the challenge to bio-medicine, the relative importance of the body and the role of the functional impairment that individuals with disabilities have is at the heart of the debates around the definition and

understanding of what disability is and how it effects people within the sociology of disability (Scambler & Scambler 2003).

2. *What are the experiences of families living with juvenile Batten's disease?*

In exploring families experiences of living with juvenile batten disease, the key findings are the effect on the family and the role of biomedicine. Juvenile Batten disease effects the entire family, not simply to the extent that they are living with an affected relative, but also through the fact that, certainly in the latter stages, the family provide the main means of maintaining the young person's self identity. The argument has been made that the families suffer the disease by proxy. The second key finding is the importance of the role that bio-medicine plays in the lives of these families, both in the control of the disease and in the provision of any future hope. Bio-medicine is not seen as the enemy but as the potential saviour. However, many of the recurrent themes within the wider literature on communication problems, power relations and information sharing, particularly around the period of diagnosis, between families and the medical profession (Ziegler 1981, Harding & Modell 1985, Schneider & Conrad 1983, Macdonald 1998) are still encountered.

3. *Do the experiences of families living with juvenile Batten disease reflect the wider experiences of people living with more common chronic illnesses?*

The experiences of families with juvenile Batten disease do broadly fit with those of people with a range of other chronic illnesses in terms of the six themes highlighted above. The main problems lie in the challenge posed by the sociology of chronic illness to bio-medicine and the social construction thesis. As already stated, bio-medicine is not negatively perceived amongst families living with juvenile Batten disease, although the actions of some practitioners may be questioned. In addition, whilst the point of diagnosis is undeniably traumatic for the parents in view of the prognosis for their child, and for the child because of the, often invasive, series of tests that they have to endure, it is not the diagnosis that creates the condition of juvenile Batten disease, as has been argued for people with a range of other, less severe, chronic conditions. The symptoms and degeneration are such that the diagnosis is a further burden rather than the main burden. The families also exhibit a distinctive coping strategy – either consciously or unconsciously – through the reduction of the disease to its immediate symptoms and their effects. The families talked about fighting one fight at a time, planning only for the next stage in the disease, and refusing to think too far into the future for fear of what it inevitably holds. In the majority of cases, therefore, the future is neither willingly thought about nor planned for. Making use of Kelly's

(1992) framework, the focus is on the technical and practical management of the condition, emotional management is rationalised through reductionism and the uncertain nature of the disease makes making sense of the condition extremely difficult.

4. ***Are the experiences of people with chronic illnesses – and in particular juvenile Batten's disease - reflected in the current literature and theory around disability?***

It seems clear that, whilst juvenile Batten disease does fit into the definitional framework of a chronic illness and clearly results in disabilities, the experiences of the families living with this disease are not adequately represented in the literature. The majority of the themes arising from the chronic illness literature are relevant, however the negative attitude towards bio-medicine is at odds with the experiences and requirements of people living with juvenile Batten disease (Scambler & Scambler 2003).

The Future of the Sociology of Chronic Illness and Disability

In seeking an answer to questions concerning the future of the sociology of chronic illness and disability, the relevance of studying the experiences of people living with rare chronic disabling conditions such as juvenile Batten disease is not always apparent. In justifying the merit of studies of this kind it is useful to reiterate the issues which arise from the experiences of the families and professionals and explore what they have taught us.

Exploring the extremes of the chronic illness and disability literatures

The extremes of juvenile Batten disease can be evidenced through the nature of the disease itself in relation to both the disease trajectory and the combination and severity of symptoms experienced. It can also be illustrated through the experiences of the families and the unique roles adopted by families living with and caring for children or young adults with this disease. The families of people with juvenile Batten disease were more than carers. They controlled all of the information about the disease, made the decisions about lifestyle, care and, towards the end, made significant contributions to the maintenance of the self-identity of the affected child. Although families are incorporated into the literature around chronic illness, this research gives an insight into the impact that having a child with a condition of this kind can have on the family unit as a whole. This will impact on our understanding of what it is like as a family living with a severely disabled child or adult.

One of the most important findings of this study relates to the maintenance of the identity of the affected child. Bury (1982) refers to the effect of chronic illnesses as 'biographical disruption'. The identity and biography of the affected person is fundamentally changed through the onset and development of the disease, requiring a rethinking of self-identity. In the latter stages of juvenile Batten disease the combination of the loss of short term memory, early onset dementia and the loss of verbal communication skills means that the affected person is unable to communicate the complexities of their new identities. Identity becomes permanently located in past experiences and preferences as the present is lost. Williams (1984) highlights the concept of narrative reconstruction in relation to the process of reconstitution of self. What appears unique in the case of juvenile batten disease is that this process of 'narrative reconstruction' is undertaken by proxy. Thus, where verbal communications skills fail, there is a reliance on the family to fill in the gaps. The family reconstructs the identity of the affected child through their past, allowing the child to make sense of their remaining long term memory through the reiteration of past events and preferences. It is the families who create the identity of the young person to the extent that it is shaped by the narrative created around the disease and the experiences of it. The affected child has knowledge of the symptoms but not the nature of the disease itself and so lacks both the knowledge and the ability to create their own illness narrative. Theories of identity have concentrated on ideas of people having a consciousness of self-identity. With juvenile Batten disease the idea of identity is much more complex – there is a proxy identity, with the crucial aspect of self-awareness missing.

Care is another area which is taken to the extremes in this condition. Children and young adults with juvenile Batten disease require specialised care. As there are few people with expertise in this area, the expertise tends to congregate in a limited number of places. This means that families will often migrate to the centres of expertise. These centres are often located within special schools or residential facilities. This means that the need for specialised care necessarily segregates young people with this disease from the wider community. Families then have to make a choice between care at home within the family environment but often without the ideal facilities, or care in a residential placement with better access to facilities but less of a role for the family. This results in a dilemma between the home and a home and between community care and ideal care. This raises interesting questions about the right of individuals to live within the community whilst receiving all of the facilities that they need, and the reality of community care in practice. It also relates to the work carried out by Voysey (1975) which identified the 'official morality' of family life within which families of disabled children are expected to function in the same way as families with non-disabled children. Thus the families face the burden of the constant need to provide care and the guilt around both failing to maintain a normal family life for non-affected siblings and the need to seek assistance from external sources when the 'constant burden' becomes too great.

The analysis of the literatures undertaken in this study suggest that both the chronic illness and disability literatures focus predominantly on the upper end

of the continuum of chronic disabling conditions. The conditions covered are relatively common and therefore easy to research. The conditions result in disabilities, but not, on the whole, in disabilities which preclude the sufferer from participating in the research. The impairments and resultant disabilities are limiting, but not to the extent that they prohibit or severely limit the agentic power of the individuals concerned. Juvenile batten disease is a rare condition, resulting in a combination of symptoms which preclude the sufferer from actively participating in the research process and severely limit their agentic powers.

Rare conditions

The rarity of juvenile Batten disease affected all aspects of daily life from the compilation of information to the nature of the expertise acquired by the families. A complex relationship between the family and the medical profession was identified. Parents had problems getting information from medical professionals, and were often dissatisfied with the communication and information received - particularly around the time of diagnosis. In addition, the rarity of the disease means that there are few doctors with any great knowledge about the condition and its impact on the child. This means that in many interactions the parents are far more knowledgeable about the specific implications of the disease than the professionals that they have been forced to consult. Again this adds to our existing knowledgeable about information sharing and the need for partnership between the family and the medical professionals.

It also adds to our understanding of the complexity of the notion of expertise in relation to people living with chronic conditions. The families in this study developed two-fold expertise as the 'expert patient' (Department of Health 2001) by proxy and as the 'medical expert'. Again this is significant as the parents are not simply armed with the experiential expertise of living with the disease on a day to day basis. The rarity of the disease, internet access and the information provided by medical professionals, scientific researchers and charities mean that the parents also develop a sophisticated knowledge base on the medical implications of the condition and its treatment drawn from medical sources. Awareness of current treatments, experimental procedures and the results of the latest research developments put the parents at the cutting edge of an area which few 'experts' are acquainted with.

The rarity of the disease also carries implications for the choice of methods in a study of this kind. Attempting to build a coherent and comprehensive picture of the experiences of people within such a limited sample population requires the use of a combination of different methods of data collection. This is a technique which would be useful for researching any condition with limited numbers of subjects, whether it is a chronic condition or a congenital or acquired disability. In addition, it was not possible, in the case of juvenile Batten disease, to collect data directly from the individuals with the condition. Therefore, all family data had to be collected without direct recourse to the affected child. This technique (although problematic if there are significant differences between the views of the proxy and those of the sufferer) could be

used to study any group of people who have profound disabilities affecting their ability to comprehend or communicate in an interview setting. Or, as in the case of juvenile Batten disease, where there are problems with the amount of knowledge an individual has about the condition they are affected by. Proxy data collection is at the heart of the methodological challenge levied by juvenile Batten disease at the social model of disability, the disease is such that outsider information is necessary if we are to have any understanding of the disease and its implications for those living with it on a day to day basis.

The ethnographic approach to multiple data collection techniques allowed for the collection of information over an extended period of time, from a variety of sources, collecting information both directly and indirectly through interview and observation. This proved successful in enabling an exploration of the key issues through the views of families, carers, teachers and doctors. In addition, the observation of the development of the Batten Disease Family Association allowed me to link the development of the group with wider developments in terms of a self-help culture and the specific attributes of different types of group. Even before an analysis of the contents of the data collected for this study was carried out, the methodological approaches to data collection challenged the existing orthodoxy in relation to both the sociology of chronic illness and disability research. The nature of juvenile Batten disease rendered user-led research impossible, and raised questions about assumptions inherent in current research around the challenge to bio-medicine and the role of the body, the social construction of chronic illness and of disability.

Embodied social actors

The reality of juvenile Batten disease poses a challenge to arguments around the social construction of both chronic illness and disability (as referenced in chapter two and explored in more detail in chapters seven and eight). The physical reality of the symptoms of juvenile Batten disease are so severe that there is no way they can be less significant than the social reality of living with the disease. In addition, there is no way that the impaired body can be separated from the lived experience of the disease. The social reality of living on a daily basis with this disease is an embodied experience which is shaped and curtailed, to some extent, by the nature and functioning of the body in which it is experienced. It is this area – encapsulated in the embodied nature of social actors – which makes the use of Critical Realism in this study particularly relevant.

The use of Critical Realism

The choices of theory and method for this study allowed for an exploration of the day to day experiences of living with juvenile Batten disease (and for an exploration of the underlying mechanisms shaping those experiences see Scambler & Scambler 2003). The frustration felt by parents attempting to get information about the disease and share information that they had already collected with less informed medical professionals could be related, for example, to power relations. In a similar way, one of the key challenges to the social model of disability arising from this study lies in the embodied nature of social life and the fact that the experience of disability encountered by the

children and young adults with juvenile Batten disease is necessarily an embodied experience and not solely a social one. The adoption of Critical Realism provided a framework through which the layers of social experience could be explored moving from the individual and the family environment, through the professional and medical spheres, whilst maintaining an understanding of the embodied nature of the disability.

The other significant advantage of the adoption of a Critical Realist framework lies in the explicit acknowledgement of an existent social reality. As outlines in the previous section, this allows us to deal with the problems posed by phenomenology and social constructionism within the sociological literatures on chronic illness and disability. A Critical Realist approach allows us to look at the experiences of families living with juvenile Batten disease on a day to day basis and at the problems that they encounter without reducing the disease to a social construction. As previously argued, juvenile Batten disease can neither be reduced to the impact of a label of chronic illness, nor can it be reduced to an inadequate social world which fails to adapt fully to the needs of disabled people. The experiences of living with this disease that families have on a day to day basis are embodied experiences bound by the inherent nature of the symptoms and disabilities caused.

Developing a critique of the chronic illness and disability literatures

There are six key gaps in existing sociological understanding of chronic illness and disability revealed by this study, relating to the account of the role of bio-medicine; the absence of an understanding of rare conditions; the need to take

into account the experiences of families of people with different types of disability; the particular issues raised by genetic conditions; the inadequate theorising of the links between the biological and the social and the methodological problems inherent in studying a rare condition such as juvenile Batten disease. The inadequacy of theorizing about the link between the biological and the social in both the chronic illness and disability literatures is addressed in the final section.

The methodological problems of studying this particular sample population have been explored in some detail in chapter four. Suffice to say that asserting the superior value of data collected from the affected person themselves, the importance of user-led research, and, in extreme cases, the dictate that only disabled researchers should carry out research on disability excludes almost all research on people with juvenile Batten disease and many other similarly experienced conditions. This exclusion would lead to and exacerbate the remaining four lacunae identified below.

There was a great deal of similarity in the experiences of families with juvenile Batten disease and people with a whole range of other more common chronic illnesses in relation to the dissatisfaction with the process of diagnosis, problems with communication with medical professionals and the need for accessible, comprehensive information. The overall view of bio-medicine in this disease, however, was one of benevolence and hope. Bio-medicine is often seen in the literature, either covertly or overtly, as the enemy of people with chronic illnesses, impotent in the fact that there are often no cures and yet

wielding power in the form of complex medical regimes and control over information and access to services. The first change that is needed, therefore is in relation to the view of the sociology of chronic illness as a challenge to the power of bio-medicine. Rather than seeing bio-medicine as the common enemy, there needs to be an acknowledgement that bio-medical research and development may be the only source of hope for people with severe degenerative conditions, and the only hope for any kind of 'normal' quality of life.

The role of the physical body and the impairments at the heart of people's experiences of disabilities also need to be taken into account. The experiences of people with severe, changing disabilities, such as those encountered in juvenile Batten disease cannot be reduced to the impact of an unequal society. It seems clear that their experiences are embodied, both constrained and shaped by the physical body through which they are forced to live social experience. Whilst a sociology of impairment, as suggested by Mike Oliver (1996) may not be the answer (see arguments in chapter eight) the findings of this study reiterate the need, as articulated by Corker and French (1999) for more research into the permeable boundary between the physical embodied experience of disability and the social experience of disability and the ways in which the two impact on one another.

The review of the literature in chapter two highlighted the gap in our understanding of chronic conditions that is created by a failure to carry out studies like this one, exploring the experiences of people with rare chronic

conditions. It would seem likely that this lacuna is, at least partly, due to difficulties of getting funding for small-scale studies with very limited sample populations. I would hope, however, that this study illustrates the importance of studies of this kind and also the value that they may have in adding to our wider understanding of this area. In addition, the absence, or scarcity of various types of disability in the literature needs to be addresses. There is the need to take account of people with different types of disability and particularly the experiences of people with profound multiple disabilities and degenerative disabilities. These groups are barely represented in the literature, if at all, and yet their experiences are often very different to those of people with less severe or static disabilities. Additionally, the number of people living with disabilities caused by chronic illnesses is set to increase with the increase in the ageing population and the improvements in acute care, and the number of people living with profound, multiple disabilities is also increasing with improved longevity amongst the most severely disabled people (ONS 2001). This will result in an increasing number of disabled people who are not adequately included, understood or represented in the sociological theory around disability.

With the increase in the number of children living into adulthood with genetic conditions, the implications of living with a genetic condition are going to become increasingly important for both the sociology of chronic illness and the sociology of disability (a point recognised in the recent work of Barnes 1999 and Shakespeare 1999), and any insight into the experiences of families living with the reality of genetic disease and the implications that this has for testing

is going to be useful. The genetic nature of the juvenile Batten disease meant that family members experiences guilt, parents felt guilty about passing on the genes that caused the disease and unaffected siblings felt guilty for not being affected. The study also illustrated the difficulties involved in making decisions about genetic testing, both prenatal and carrier testing, and decisions on whether to try for more children, whether or not to have any future children tested and what to do if tests come back positive.

It would seem, from the above, that studying the experiences of families with juvenile Batten disease has added to our understanding of the wider issues around chronic illness and disability. The implications of this new information for future developments within the field of chronic illness and disability is discussed in the concluding section with reference to seeking an answer to the final research question.

New Paradigms, New Horizons

The summation of this thesis comes in the form of the answer to the final research question asked at the start of the study:

> **Is it necessary to develop an alternative approach to the theorising of chronic illness and disability to reflect the experiences of the many groups of people with rare chronic conditions who do not appear to be covered in the existing approach?**

Having illustrated the convergence and divergence between the experiences of families with juvenile Batten disease and the wider sociological literatures on chronic illness and disability it seems clear that there is a need for a rethink in the way that sociology approaches the study of these areas. Certainly there is room for the study of rare conditions such as juvenile Batten disease as it seems clear that this can both add to our existing understanding and give us a new insight into living with chronic disabling conditions in the twenty-first century.

Current theories about chronic illness and disability are inadequate in that the body and the social are not linked. It is within this key area that this study is most important. The phenomenological and social constructionist approaches outlined in some detail in chapter two suggest that it is the labelling or the social environment which are the predominant issues when looking at chronic illnesses or disability. Although there is a growing awareness of the need to reintegrate the body, predomiantly through the work of Shakespeare (1999) and Corker and French (1999), it remains unclear as to exactly how this integration is proposed. This study suggests a framework through which it is possible to both accept the inherently embodied nature of social action whilst also allowing for the experiences of the families to shape and be shaped by the social world.

The use of a Critical Realist framework allows for acceptance of the body as a generative mechanism in the realm of the real. In the words of Sayer (2000) the real refers to the structures and power that lies within objects:

> 'Whether they be physical, like minerals, or social, like bureaucracies, , they have certain structures and causal powers, that is, capacities to behave in particular ways, and causal liabilities or passive powers, that is, specific susceptibilities to certain kinds of change.' (Sayer 2000: 11)

Thus the body is a physical structure which is constrained by the power to work only in certain ways. If viewed in this way the embodied nature of social actors means that social action is constrained by the nature and functioning capacity of the body that embodies it. When the ability to study the social world at the levels of the 'Empirical' and 'Actual' are added to this we have a framework through which we are able to link the social and biological without losing the essential importance of either aspect within out understanding of chronic illness and disability.

In addition to the need to link the biological and social I would suggest that there is a need to widen the remit of the sociology of disability to acknowledge the embodied nature of disability and better incorporate disabilities caused by chronic illnesses, those caused by rare diseases, profound multiple disabilities and degenerative disabilities. There is a small body within the field of disability theory who, whilst demonstrating that they are able academics who happen to have disabilities, and yet who are clearly shaped by their disabilities and gain a deeper understanding because of them, appear to disregard the group of people with more severe disabilities for whom this is not possible.

Associating with profoundly disabled people, as with older people, chronically ill people or those with learning disabilities, actually attacks the arguments being made for all disabled people to engage in agentic activity. I would suggest the need either for a separate body of work looking at degenerative or chronic disabilities to be incorporated into the disability literature or, ideally, a middle ground incorporating chronic illness and the associated disabilities within the wider understanding of disability. This would seem the best option because all chronic illnesses cause some degree of disability or impairment, whether this is or is not visible and apparent, which curtails or reshapes the life of the affected person to a greater or lesser extent. Therefore, combining the two literatures would enrich both and ensure that the experiences of the significant number of people with disabilities caused by chronic illnesses are fully included in debates around definitions, political action and civil and human rights.

Appendix 1

Heather House

I was initially involved in research looking at the social care needs of young adults with juvenile Batten disease with the remit of developing a philosophy of care for a new care/nursing home built specifically to cater to the needs of this group and others with similar sets of symptoms and life expectancy.

Heather House is a nursing and social care service for young adults with a visual impairment and profound multiple disabilities or degenerative conditions such as juvenile Batten's disease, which result in a shortened life expectancy. The residential accommodation consists of two linked bungalows. Each bungalow contains eight ensuite bedrooms, two communal bathrooms and a communal living, dining and kitchen area. There is also an activity and resource centre which contains a large physical activity hall, hydrotherapy pool, multi-sensory room and a variety of activity rooms in which everything from trampolining to computer studies can be offered. The purpose-built development offers:

- A Nursing and Social Care Service for 16 young adults (aged 18-35) with a visual impairment and a degenerative illness with a short life expectancy, especially juvenile Batten disease.

- An Activity and Resource Centre offering a range of services to the residents of Heather House and people living in the community.
- A Family/Carers support service that will include sharing ideas, concerns and issues.
- An Outreach Service to support people with a visual impairment living in the community and to offer visual impairment expertise to other professionals.
- On-going research in the area of degenerative illnesses affecting young people. (taken from Seeability's brochure on Heather House, p2).

The aim of Heather House is to provide an environment which focuses on maximising quality of life, run on a holistic basis and incorporating a whole range of therapies and expertise within the multi-disciplinary care team. The homelike environment is young, lively and relaxed, reflecting the needs of the people who will be living there and the day to day care is flexible adapting to the changing needs of the young people. Emphasis will be placed on inclusion and the young people will be facilitated in taking part in all activities whether actively or passively. Heather House has been purpose designed and built for people with a visual impairment, incorporating colour contrast and tactile features in all areas. In addition to this all areas of the site are large enough to accommodate beds so that the young people who need to spend considerable amounts of time in bed are able to access all areas and all activities. Heather House provides a service for young people right through to terminal care and emphasis is placed on the importance of

the role of families in the care of their loved ones. It is a place where the focus is very much on living and quality of life.

Appendix 2 Batten Disease Family Association Newsletters

Attached are the first three newsletters produced by the Batten Disease Family Association. This gives a good idea of what the group is about and how it has developed since its inception at the end of 1997.

Batten Disease Family Association Newsletter

March 1999 Issue 1

Welcome to our first BDFA newsletter! The main purpose of this letter is to let everyone know that we now have a family support group for Batten's disease for ourselves in the UK.

The principal aim of the group is to provide contact and support to families affected by Batten's disease. We intend to keep families informed about recent developments and research at a national and world-wide level. We hope to raise awareness of the illness and hopefully in the future to be able to raise funds to facilitate further advances in understanding and perhaps one-day treating Batten's disease.

The Story So Far

The first step was the conference in June 1997 organised by Michelle Bazeley and her colleagues at St Andrews in Scotland. This brought together people from seeAbility (particularly David Ireland & Joan Deeley) and interested families who saw the need for an ongoing group and conference-like get togethers. seeAbility have become particularly involved because of their interest in helping young adults with Juvenile Batten disease, this has lead to the development of Heather House - a purpose built residential care facility to meet the needs of such people. After the conference Sasha Scambler was given a research post at seeAbility and began contacting families to collect information about juvenile Batten disease.

seeAbility together with a group of parents (the Newcombes, the Shingles and the Daniels) began to plan and organize a conference at Warwick University for the summer of 1998 with the wish to discuss the setting up of an association but sadly it didn't come to fruition.

Instead a meeting of eleven or so families was held in November 1998 at the Society for the Blind Resource Centre in Leicester. The meeting was facilitated by seeAbility, and introduced by David Ireland. With the help and guidance of Linda Partridge of Contact a Family it was decided with enthusiasm that we very much needed and wanted a support group.

A few volunteers stepped forward to form a steering committee and the first meeting of this group was held in February. We covered quite a lot of issues - here's a summary .....

- We have a name as you can see, we are the Batten Disease Family Association
- seeAbility have been very supportive in many ways and have offered us the use of their new residential facility at Heather House, Heather Drive, Tadley, Hampshire RG26 4 QR. which is due to open in the early summer, as a permanent postal address for our association. Until it opens, post should be sent to the

Leatherhead address as given later.

- We decided on a Mission Statement to sum up what BDFA represents

> *...to provide a supportive, informative, networking organisation for the families, carers and professionals giving care to children and adults with Batten disease*

- Individual roles within the steering group were decided

Chairman:	Irena Newcombe
Secretary:	Pauline Muncey
Treasurer:	Sharon Burnham
Newsletter Editor / Medical liaison:	Chris Martland
Committee members:	Jim Docherty, Cliff Burnham
Co-opted member:	Sasha Scambler

- **Funding**; it was felt that to cover basic running costs, we will need a subscription system which will be voluntary. See the membership application form later.
- **Logo**; we need a logo, see competition later!
- **Web page**; we felt this was important and plans to develop a site are being looked into.
- **Communication with other groups and agencies**; we plan to contact allied associations such as BDSRA in the USA, the RTMDC, the Weymouth BRST and Contact a Family to formally inform them of our existence and about the BDFA. We also intend to create and distribute a leaflet about the BDFA and Batten's disease to other charities, paediatric and neurology departments, special schools etc to both advertise our presence and raise awareness of the disease.
- **Register of families**; we only have a small database of families with Batten's disease, this was mainly made up of people who went to Michelle Bazeley's conference in St Andrews. seeAbility and the Department of Paediatrics, University College London (UCL) who you may already know are setting up a European register also have lists but because of confidentiality concerns, we at BDFA do not have access to these lists. To create a more accurate comprehensive register, we need to know who you are! To achieve this, we need you to return the completed membership form. This newsletter may have come to you via seeAbility or the UCL, so until you return the form, we may not be aware of you at BDFA. I hope that makes sense!
- **Family Networking**; one of the reasons for setting up an association is so that we can all be there to support each other, with each family's consent we would like to create a network so that we know who and where other families are together with contact numbers etc. We understand that not everyone may wish to have such information publicly available (i.e. in the BDFA newsletter) - the membership form has an opt out option.
- **Next steps**; The steering group meet next at seeAbility's headquarters at

Leatherhead on March 20th, we will discussing the issue of becoming a registered charity which would include the drawing up of a formal constitution, we will also be looking at the up and coming conferences. We will also be discussing whether to have a Patron as a figurehead for the association.

If anyone wishes to become involved with the steering group, or has any ideas to discuss, please get in touch with Irena.

The Newsletter; This and probably the next one will be in the form of a progress bulletin to let you all know what's happening. I hope soon to put together something a bit more attractive and family-friendly. I can't do this on my own - *I will need contributions*! I would like to put in features about ourselves, our children and experiences, articles about carers and what help is available, items on practical issues such as benefits, education, aids & appliances etc, medical features looking at medication, feeding problems, tubes etc, science updates on recent developments and progress. I would like to also have features for siblings - written by sibs for sibs.
Please start writing; life stories, a day in the life type articles, descriptions of special carers or agencies that warrant praise, write about difficulties and problems (we shouldn't find that too difficult), poems, uplifting quotes. Maybe even cartoons! Whatever, send them in. It is your Newsletter - for you. You will make it what it will be!

Send items by post to :
BDFA Newsletter Editor
c/o seeAbility
56-66 Highlands Road, Leatherhead, Surrey, KT22 8NR
or electronically by e-mail to
cpm@cmartland.freeserve.co.uk

Logo Competition

Calling all artists! Have a go at designing a logo for the BDFA, something that in an image conveys something of what we are all about.
Send your entries by post to:
BDFA Logo Competition
c/o seeAbility
56-66 Highlands Road, Leatherhead, Surrey, KT22 8NR

The winner will be chosen by the steering group and the winner will receive a Marks & Spencer voucher for £15, good luck, start drawing.

Conference News

seeAbility have two events on the agenda over the next few months. Firstly there will be a conference on June 30th 1999 aimed mainly for professionals but to which families are invited.
Secondly, it is hoped that we will have a Family Conference at the end September / early October. This will be held at Heather House on a Saturday and will consist of a day of lectures / presentations and hopefully some kind of social event in the evening. I hope to have more information in the next newsletter on this. If you have any thoughts on topics or speakers for the lectures, please let Irena know ASAP.

The Steering Group - *Who are we?*

Irena Newcombe My husband David, and I have two children . Alex, our son, is 15 years old and Helena is 11years old. We received the news that Helena was affected by Juvenile Batten disease on 6th March 1997. Since that date I have been

grateful for the continuing contact, knowledge, support and friendship shared with and by other families and organisations, both here in the UK and abroad.
I feel passionately about the potential benefits of The Batten Disease Family Association and look forward in playing a part in its future success.

Sharon & Cliff Burnham : Sharon is 39 years old, and works as a project manager for BT in London. Cliff is 44 and works as a Quality Liaison Officer for BT in London. We have two children, Lucy , age 11 and Katie age 8. Lucy was diagnosed with Juvenile Battens Disease 3 years ago. Sharon is secretary of a local charity called VIC, supporting Visually Impaired Children and their families and has recently become a Named Person for the RNIB.

Pauline Muncey I live with my two children, who both have Juvenile Batten Disease, in Crowborough, East Sussex. Both Joanna (aged 14) and James (aged 9) go to Dorton House School in Sevenoaks and I work full-time as an I.T. Project Leader. Jo was diagnosed nearly 4 years ago and James soon after. We try to live as 'normal' a life as possible (whatever normal is!!) and have fun!!!!'

Jim Docherty: I am a Project Manager in the construction industry and live on the edge of Ashdown forest in a little village in East Sussex called Nutley. I have 3 children, ages ranging from 24 to 6. My connection with Batten Disease is through my friendship with Pauline, Joanna and James.

Chris Martland I am a 36 year old GP in Huddersfield, West Yorkshire. I am married to Janice and have two children; Charlie age 7 and Maisie age 5. Charlie was diagnosed with Late Infantile Batten's disease 3 years ago.

Sasha Scambler I am a medical sociologist working for seeAbility, a charity providing residential and day services for adults with a visual impairment and other disabilities. I am currently researching into the social care needs of children and adults with juvenile Batten's disease and have been involved in the setting up of the BDFA and in compiling information on the juvenile version of Batten's disease.

Contacting the BDFA

By e mail BDFA98@hotmail.com
By post
(Temporarily)
BDFA c/o
56-66 Highlands Road, Leatherhead, Surrey, KT22 8NR
in the near future this will become:
BDFA c/o
Heather House, Heather Drive, Tadley, Hampshire RG26 4 QR

Steering Group

Irena Tel 01636 708028
Email newcombe@clara.net

Sharon + Cliff
Tel 0181 896 2401 evenings
Email sharon.burnham@bt.com

Pauline + Jim Tel 01892 653604
Email
Pauline.Muncey@pppgroup.co.uk

Chris Martland tel 01484 660386
Email
cpm@cmartland.freeserve.co.uk

Membership form

Family Registration Details

This information will be held confidential within the Batten Disease Family Association and will be covered by the Data Protection Act

Surname _____

Parents first names _____ _____

Children: Firstnames and dates of birth _____

_____

_____

_____

Please indicate which children have Batten's disease and if possible which form of the illness eg Late Infantile, Juvenile etc

Address _____

_____

_____

_____

Postcode _____

Phone _____

Email _____

I wish to become a member of the Batten's Disease Family Association and receive the Newsletter

I am able to enclose £____ for the annual membership subscription fee

(Suggested amount £10 for families but this is entirely voluntary)

For professional subscription £40

Consent for family networking

I am / am not * happy for the above information to be made available to other members of the BDFA

* delete as appropriate

Signature _____ Date _____

Please send the form to BDFA c/o seeAbility 56-66 Highlands Road, Leatherhead, Surrey, KT22 8NR
Please make cheques payable to Batten Disease Family Association.

Batten Disease Family Association Newsletter

Heather House, Heather Drive, Tadley, Hampshire RG26 4QR
http://www.bdfauk.freeserve.co.uk BDFA98@hotmail.com

July 1999 Issue 2

Hello again! Here's the second "progress report" newsletter to update you on what's happening and what's in the pipeline.

Thankyou to all of you who have returned your membership forms (and kindly your subscriptions).

If you haven't sent your form in, please do, so that we can continue to send the newsletter to you. This is the last newsletter we will be able to send to you unless we have received your form.

I would like to say thank you to all the people - from parents to professionals who have given encouragement and wished us well with the Association.

We had the second meeting of the Steering Group on 20th March at seeAbility's headquarters in Leatherhead and were very pleased to welcome Simon Carter, Christine Foster and Andrew Henery.

We discussed distribution of the newsletter and a leaflet (still under construction), and hope by these means to get information about Batten disease and the BDFA far and wide. Not only to ourselves as families, but to children's hospices, paediatric, neurology and ophthalmic clinics, special schools and other associated charities and parent support groups, such as the RTMDC.

I am also writing to the journals of those professionals who help us care for our children - e.g. physiotherapy, OT and nursing.

At our recent meeting, we were joined by Linda Partridge from Contact-a-Family. We had a lengthy discussion about the pros and cons of becoming a registered charity. The main advantage would be that people would take the Association a great deal more seriously particularly with regard to fund raising. There are some legal and bureaucratic obstacles for us to work through, but we are fairly confident that if we do apply that we would be successful. As a group, we were unanimous in wishing to proceed with the application, but we would like to put the matter to you as members. So please find enclosed with this newsletter a voting paper to let us know how you feel about becoming a charity.

We have some exciting news to pass on. We have details of a patron for the Association, news about our web site address and a report on the recent conference on Batten disease - "Care in Partnership". There is a new book about Batten disease written by Dr Sara Mole in London, and a very moving personal account of living with this dreadful illness from the mother of a little boy with Infantile Batten disease.

Chris Martland Newsletter Editor

News

We are pleased to announce that we have a patron for the BDFA; **Terry Gilliam** of Monty Python fame. Terry was the creator of the Python zany cartoons and animations, he has also directed films including; Adventures of Baron Münchausen, Brazil, Jabberwocky, The Fisher King, Time Bandits, Monty Python and the Holy Grail and 12 Monkeys. Terry has become aware of Batten disease through friendship with Andrew Henery & Christine Foster whose son Matthew has late infantile Batten disease.

Batten Disease Conference - June 30th 1999

This was not only the first conference that the SeeAbility and the BDFA have organised but also the first to be held at the new facility at Heather House. The focus for the day was on "Care in Partnership", with delegates and speakers from all backgrounds (including professionals, carers and parents) coming together to share their expertise and experiences.

Joan Deeley, from seeAbility and the project manager for Heather House opened the conference with a warm welcome to all. Starting the busy programme, we heard from Dr. Sara Mole, from University College London (UCL). Dr. Mole outlined the work undertaken by the team, initially identifying the gene for Juvenile Batten Disease and subsequently studying Late Infantile Batten Disease. We were given an update on the genetics and an idea of the complexity and difficulties involved in trying to find the mutations that bring about the different types of Batten disease.

After a break, Dr. Paula Sneath discussed the doctor's role in a multidisciplinary team. Dr. Sneath has been an Honorary Paediatrician at the Children's Trust in Tadworth and the doctor responsible for young people with Batten disease since 1995. She described the problems encountered from the doctor's point of view in the progression of the Batten illness, and her ways of approaching and trying to deal with these issues particularly with regard to medication.

Sasha Scambler has worked for seeAbility since 1997 as a Research Officer looking into the social care needs of young people with Juvenile Batten Disease. She is part of our Steering Group and has been greatly involved in the setting up of the BDFA. Sasha described how her work initially involved collecting as much information and literature that was available, contacting health authorities, special schools, advisory services, genetic units, and a range of help groups. Meetings followed with paediatricians and interviews with families who had children or young people with Juvenile Batten disease. The recommendations made in her report were used when planning the excellent facilities and care packages at Heather House.

We then had lunch, during which delegates had chance to "mingle" and chat.

In the afternoon we heard from Dr. Ruth Williams, who now works part time as a senior registrar in Paediatric Neurology at Great Ormond Street Hospital. Dr. Williams also spent 5 years prior to this undertaking research in Professor Mark Gardiner's laboratories at University College London (We are also pleased that she has agreed to be Medical Advisor for our association). Firstly, Dr. Williams spoke about the clinical aspects concerning diagnosis of all the types of Batten disease, and the management of developing needs. We then heard how, with the help of a grant from the European Community, work started in 1996 to set up a European Concerted Action Database for NCL (Batten Disease). The aim was to get European scientists and clinicians together to talk to each other, share information and to develop a database of clinical resources which could be used throughout Europe.
Dr. Williams explained that the long-term objectives of such a database included collecting epidemiological data, i.e. how many children and young people were affected, and to explore relationships between actual genetic mutations and the clinical appearance of the disease. This information is not only helpful when trying to evaluate and plan for services in health, education and respite care that will be needed for these children, but also as a baseline for possible future therapeutic trials.

The delegates then divided into two groups to attend workshops, which were facilitated by Peter Mannering and Sarah Kenrick. Peter is a teacher from Dorton House School for visually impaired children in Sevenoaks, and Sarah worked at The Children's Trust in Tadworth until recently when she became the manager of Heather House. In these workshops we had the opportunity to discuss "Care in Partnership".

The final summary session included feedback from the workshops and comments and questions for the panel. During this discussion the need for some sort of knowledge base was identified, although differences have been experienced geographically, it was felt that it would be useful to have an overview of services and expertise available.

I felt the seminar was very interesting and well presented by the speakers. I was encouraged that there are areas of good practice of partnership in care and hope that with the help of the developing BDFA these will grow.

Irena Newcombe

Batten Workshop in Bethesda, Maryland USA

In April there was a conference to look at future developments in Batten disease research. A summary is being written which will be posted on the web site when available and probably included in the next newsletter. Many of us have become aware of some potentially exciting work on enzyme replacement therapy for the classic forms of infantile and late infantile Batten disease, but it is important to remember that this work is in its very early stages and that there is a long

way to go yet before any of the scientists could be confident of being able to slow down or treat this illness.

New Book on Batten Disease

Dr Sara E Mole PhD, Senior Lecturer in Molecular Genetics at University College London has been involved with the writing of a new book on Batten Disease. The book is called *The Neuronal Ceroid Lipofuscinoses (Batten Disease)*, Publisher IOS Press, and price £86. I asked Dr Mole to outline what the book is about, she replied ....

..... In 1996 twenty-five groups in Europe with an interest in Batten disease were brought together under a European Union grant co-ordinated by Prof Hans Goebel. In the 3-year lifetime of this "Concerted Action" the participants met regularly to consider and concentrate on different aspects of Batten disease. At the end of this period a Monograph was written by the participants to summarise the current state of knowledge of Batten disease. This is now available as a 211-page book covering all aspects of Batten disease. There are 18 chapters, a comprehensive list of references and an appendix listing Batten support groups and other useful addresses throughout the world.

As well as an introductory chapter there are also chapters defining the ultrastructural patterns observed in patients' cells, and the consideration of diagnosis using morphology. A chapter that will be very useful to clinicians is the set of diagnostic algorithms developed by those experienced in diagnosis of Batten disease. There are individual chapters on each genetic type (CLN1, CLN2, CLN3, CLN4, CLN5, CLN6, CLN7, CLN8 and remaining variants). These chapters, where appropriate, start with the clinical details including diagnosis and patient management, progress through the morphology associated with that type, and conclude with up to date results arising from research in the fields of molecular genetics and cell biology. Each of these chapters is accompanied by a comprehensive set of figures. Additional chapters include an up to date review of animal models for Batten disease and the consideration of Batten disease types in 13 different European countries. There is also a chapter summarising the disease-causing mutations known to date (60 over 4 genes). The book closes with an outlook into the next decade.

The book was designed for clinicians involved in diagnosis and for clinical and non-clinical scientists engaged in research into Batten disease. It will also be helpful to those involved in the care of patients.

Dr Mole has kindly donated a copy of the book to the BDFA for reference and this will be available to members to borrow.

Sharon Burnham, who has an 11 year old daughter with Juvenile Batten disease, reviewed the book from a parent's perspective, here are her observations.....

"On a recent visit to Heather House I was lucky enough to collect a copy of the book. This is the first of its kind, a medical bible on Batten Disease. It contains contributions from specialists from all over the world who have been involved with children and adults suffering from

Batten Disease. When I first started to browse through the pages, I must confess to being overwhelmed by the medical terminology, references to genetics and illustrations of curvilinear profiles! As a parent and not a doctor, I suppose this was only to be expected. However on closer examination, excuse the pun, there were specific areas of the book that were interesting, informative, and encouraging.
The book is divided into the variants of the disease, and within each variant it gives background, history, symptoms, medical care, some research and findings. My daughter has Juvenile Batten Disease and what I found particularly useful in this section was the reference to what medical care should be available, as well as going into greater detail about the symptoms. If as a parent you are interested in the genetics and medical side of the illness then I am sure this book will be of great interest to you. If you are seeking a more practical guide to caring and managing the illness then I am not sure you will find what you are looking for in here.
However, let's celebrate this achievement, a book, and the first of its kind, about the illness that we all know only too well. Lets hope, that, with this information at the finger tips of both the medical profession and of ourselves, we can continue to strive for the best care for our children and a cure to this debilitating disease."

I also have had a chance to read the book and although as Sharon says - it is quite scientific it is obviously written not for families and parents but for the geneticists, neurologists, paediatricians and researchers involved with Batten disease. I feel that this book is a very important piece of reference material and the authors, editors and publishers deserve praise and congratulations.

Blood Test for LINCL

The CLN2 enzyme assay is now available as a diagnostic test at Great Ormond Street Hospital for Children. It will be most valuable for use in those children who have a history suggestive of late infantile Batten disease, but could be used as a pre-symptomatic or prenatal test with or without skin biopsy and genetic testing. **This would be particularly useful in young siblings of affected children before any signs of the disease were present - the child could have a blood test instead of requiring a skin biopsy.** If you would like to go ahead with a test, your GP or paediatrician should be able to organise this with the following advice. *The test is done on a blood sample (5mls LiHep) which can be sent to the Enzyme Laboratory, Camelia Botnar Laboratories at the Great Ormond Street Hospital for Children, Great Ormond St, London WC1N 3JH. The samples should be sent to arrive within 24 hours and not on a Friday. Professional enquiries should be directed to Dr Young, Chief Biochemist.* For any other information or enquiries, please contact Dr Ruth Williams Department of Paediatrics, UCLMS, The Rayne Institute, University Street, London WC1E 6JJ

Web Site

We have ventured boldly into cyberspace! Alex Newcombe (Irena's son) has put together a web site which has lots of information on

Batten disease and the BDFA. It also has links to other interesting sites.
If you have Internet access check it out at http://www.bdfauk.freeserve.co.uk
Well done Alex!

Conference News

The family conference in October is definitely on! It will be at Heather House on Saturday 2nd October.
SeeAbility will be helping us to host the event. There will be staff on hand to care for the children. We hope to start at 10am and have a morning of presentations including:

- Dr Irene Hofman (Consultant Paediatrician, Netherlands) speaking on the Medical management of Batten Disease
- Dr Hannah Mitchison (Lecturer in Molecular Genetics at UCL) hopefully bringing us up to date with developments in genetics
- A BDFA update. This will allow us to introduce ourselves and to share ideas about how we would like the Association to develop.
- After lunch, we would like to have a session "Social Care" focusing on accessing and getting the best from social services, education services and benefits.

We expect the conference to finish at about 5.00pm but there will be plenty of time and opportunity to chat and meet the speakers and other parents etc.
We really hope that as many people as possible will make the effort to come together.

SeeAbility are sending out application forms, if you don't receive one within a week of this newsletter, please ring: 01372 373086 or Fax: 01372 370143

Closing date for completed applications forms: Friday 3rd September 1999

Heather House

Heather House is a nursing and social care service for young adults with a visual impairment and profound multiple disabilities or degenerative conditions such as juvenile Batten's disease, which result in a shortened life expectancy. The residential accommodation consists of two linked bungalows. Each bungalow contains eight ensuite bedrooms, two communal bathrooms and a communal living, dining and kitchen area. There is also an activity and resource centre which contains a large physical activity hall, hydrotherapy pool, multi-sensory room and a variety of activity rooms in which everything from trampolining to computer studies can be offered.

The aim of Heather House is to provide an environment which focuses on maximising quality of life, run on a holistic basis and incorporating a whole range of therapies and expertise within the multi-disciplinary care team. The homelike environment is young, lively and relaxed, reflecting the needs of the people who will be living there and the day to day care is flexible adapting to the changing needs of the young people. Emphasis will be placed on inclusion and the young people will be facilitated in taking part in all activities whether actively or passively. Heather House has been purpose designed and built for people

with a visual impairment, incorporating colour contrast and tactile features in all areas. In addition to this all areas of the site are large enough to accommodate beds so that the young people who need to spend considerable amounts of time in bed are able to access all areas and all activities. Heather House provides a service for young people right through to terminal care and emphasis is placed on the importance of the role of families in the care of their loved ones. It is a place where the focus is very much on living and quality of life.

If you would like more information about Heather House please contact Sue Ogden, Placements Co-ordinator, at: seeAbility, 56-66 Highlands Road, Leatherhead, Surrey, KT22 8NR. Tel: (01372) 373086 or e-mail: S.Ogden@seeability.org. Alternatively visit our web site at: www.seeability.org

Families up close

As I mentioned in the last newsletter, I hope to include features about our lives our children and ourselves. The following article is in the form of a diary - put together by Mrs Fran Theobald. Her son Jay who is 2 has infantile Batten disease, she also has a 6 month old daughter called Tianna as well as two older children, the family live in Colchester.

[When I first read this, I thought it ought to be edited down as it is quite long but I felt that it says so much about the day in - day out worries and difficulties that we all have to try to cope with that I have left it unabridged - Ed]

15th January 1999

From the day Jay was born I felt something was wrong with my baby even though he presented himself to be completely normal.

He smiled and cooed and progressed through the milestones up until about 7 months when I noticed that he still showed no signs of sitting up. This obviously concerned me but I was told not to worry as he still had time to pick up, eventually he sat up at approx. 9- 10 months. He did not start to get mobile until 11 months and this was very slow to take off. He showed no signs of trying to stand. He was always on antibiotics for his chest and was asthmatic. We were referred to the consultant for the flatness of the back of his head only to be told this is a normal thing due to sleeping on his head all the time. Again we were sent home.

Milestones got no further and after many visits to the hospital including being told that I was an overreacting mother, the doctors eventually were beginning to listen to my concerns. Jay could say many words and played like a normal child, but I noticed he had started to forget them then they would return. Although I told the doctors that he was losing skills, we were told it was probably developmental delay. He had regular physio and occupational therapy with no improvement. At last we were referred to a neurologist who showed great concern and had Jay up to Addenbrookes for an M.R.I. scan and blood tests.

In the meantime Jay lost all his speech and the ability to play - his only pleasure was the Teletubbies, he was fascinated by the lights on the stereo and the washing machine. He screamed all day and never laughed, bouncing him on your lap sometimes produced a little smile - a treasured moment. Xmas came and went and waiting for Jay's results was horrendous - the not knowing. Jay became harder to care for he just

screamed and moaned all day and there wasn't a thing we could do to help him.
Eventually a phone call came, a possible diagnosis, I felt a glimmer of hope, but however you try to prepare yourself for possible bad news, nothing can prepare you for what we were told - Jay has got Battens disease. At this stage, it wasn't 100% diagnosed but they were pretty confident. We had to go back to hospital for a skin biopsy just to confirm it on paper I guess.
We have to sit and watch our baby boy die in front of us and there's nothing we can do to help him he will slip into a vegetative state and not know that his family is there. The children are devastated, how do you tell siblings such a terrible thing? All we can do is cry and wait for him to die, the pain is so unbearable and you feel so alone and so helpless then you fear for your younger ones.
I speak as though Jay has left us already because it's how it feels at this point. The numbness and hurt dig like a knife, you feel like nothing matters anymore but it does, the children matter, you have to cope, you don't have time to think for yourself when you have others to care for, and that can be hard too. As time goes by I will fill in this horror story, all we have is a small memory of our darling little boy who laughed and played. I keep telling myself, it's not Jay we see any more and I try to hang on to the happy memories of him previous to this cruel disease. I hope that when all this ends and we can begin to grieve, then to fight back, that this will help other parents to realise that they are not alone, and that you must carry on for any of the other children, as hard as it may seem. I shall end at present on this note until Jay goes through another change so that this may help who ever reads it.....

16th Jan 99
Jay had a good night last night we took him to Tesco to take the kids minds off the situation. He was reasonably happy as he was quiet, then he came home for a nap. In the afternoon he was very unhappy, he cried and moaned for ages, he ate his tea - well I'm so glad we can still feed him I cried a lot today but it hasn't helped at all.
We all kept busy today trying to be as normal as possible the kids seem to keep you going. Mum is coming back to England on Thursday to be with us - I don't want her to be here at the later stages of Jay's illness, I want everyone to remember him as he was. I feel as though I want to protect the rest of the family, it's enough with us having to cope with it. They all cried on me - I feel as though I have to support everyone else. If I hear "I'm sorry" once more I shall scream I'm sure. I know they all mean well but it makes it so hard to deal with. The hospice rang today, we're going to visit on Friday they were so kind I felt that I they were close friends already - before I'd even met them which is a great comfort. I know we have a long haul ahead of us and my fear for Tianna is growing day by day. [Tianna is about 6 months old and has not been tested for the disease - Ed]
Jay is a bit quieter now he loves to watch the Teletubbies, I feel we owe them a lot too, his room is full of them right up to the ceiling, thanks to two very dear friends Sharon and Ian who I could not do without. Jay has a very special bond with them too, since they started to foster him to give us respite which is needed with these cases or you would go mad, as you feel so helpless. It seems as

though Jay has touched many hearts around him as though maybe he was too good for this world, it's like this with many sick and dying children, I'm sure there is a better place for them somewhere. I'm not a religious person neither is my husband but I'm sure there is something somewhere for such precious little people.
Well another day tomorrow I expect I shall cope, I guess I have to even though I feel like my whole world has crumbled around me and it feels like nothing matters anymore, time is a great healer they say, we shall see I suppose.....

19th Jan 99
Today we had a fantastic response from Jay, he actually smiled! I know we won't see that much more so it was a treasured moment for us all to see. The children thought it was the greatest thing ever, it brought tears to my eyes, a moment I will never forget. He had a bad night and had to have chloral, which made him a bit sickly today. He has been unhappy this evening though - I'm sure something is going on. Social services took him to Stepping Stones today he seemed to enjoy being with all the other children. Everyone has been so kind, they are trying to raise the money to fly us all to Spain to spend time with nanny and grandad, I do hope it goes ahead as this will be good for the others to do something special with their little brother.

21st Jan 99
Jay has gone out with Sharon to see Ian today it seems so quiet in the house. He seemed a bit upset before breakfast, but when Sharon phoned me she said he was fine, and had fallen asleep in her arms. I must admit he usually is happier when he goes out in the buggy. We are picking mum up tonight, I cant wait to see her it's amazing how this can bring a family so much closer, I only wish she was coming for a better reason. Steve has been great he has held us together and we are so close now, I'm so glad this has not destroyed us, as it does in so many other families. The one thing I have learnt from all that we are going through is that you must hold on together and help each other through this however hard it may seem.

6th Feb 99
So much has happened in the past few weeks. Mum came home and we all visited the hospice, which is a lovely place and all the children wanted to stay, even Jay had a nice time, his face lit up in the hydrotherapy pool. During the next visit we shall go out, as I feel confident enough to leave him with the nurses. We have had a really nasty few days Jay went into a stage of non stop screaming there was nothing I or Sharon could do to calm him. He stopped eating, I called the nurse and she gave him a nasogastric tube. This made him worse, I felt so helpless that the nurse advised me to take him to the hospital for a period of respite. As much as I hated doing this I had to and I'm glad I did, the staff were great, at least I knew he was safe. A junior doctor saw him he had no idea and was very rude. The next day a senior doctor saw him and his drugs were reviewed; clonazepam 5ml twice a day and 2.5ml twice daily if he needed it also diazepam if he starts to get bad again. At last he settled they kept him in for a few days on a high energy feed and he began to feed off the spoon again, I've no idea how long for but fingers crossed. Later on he was violently sick and had diarrhoea but they let

him come home and he hasn't kept his dinner down at all so we shall monitor him.
The kids took it all very badly but there's not a lot you can do, you feel so useless. The newspaper did an article to try to raise money to go to see nana and grandad with Tracie's help and everyone else it may take off. The Dream Team have stepped in and they seem to be able to achieve their goal, any way we shall have to see, so watch this space, I'm so grateful for all that is being done to help improve my little boy's short life.

13th Feb 99
Jay is still in the hospital with a stomach bug, his temperature has gone from very low to high - it's so erratic. His heart rate dropped quite low at one point and I found myself asking him to go and end all the suffering as cruel as it sounds. I'm sure any parent in our situation would be doing the same. Steve and I have been up and down the hospital like yo-yos. I couldn't go for four days as I got the flu and a chest infection and I didn't want to pass it to Jay as I would feel so guilty if I had given him any thing to make him feel more poorly than he already does. I'm coping well at the moment but I know when the time comes that the story will be different. You have to deal with one day at a time I'm told, but I have to be there for the rest of the family too. Steve's finding it very hard, we do argue occasionally but deep down I know that we will get through this one way or another. Well tomorrow I hope Jay will be ok, we shall see bless his little heart.

21st Feb 99
Well Jay has come home, the first days were bad but he is a bit better now he is on clonazepam and diazepam, chloral hydrate and a nasogastric tube which I've got used to now, he has deteriorated a lot more but he is just about sitting up still. The Dream Team are doing ok as far as I know and we've been in the Gazette and Anglian Times, and on the radio - all a bit daunting but if it gets us all to Spain to see mum and Sid then I will keep on with the fund raising. Back to the hospice this week, I'm looking forward to it in some respects not in others, we spoke to Chris and Andy Marks on the phone last week it was good to talk to someone who has been through this awful battle. They are lovely people and I feel I've known Chris for years. Well, we must battle on and live in hope for little Tianna, I can't do this again but I know I may have to - this scares the hell out of me.

5th Mar 99
Jay has been back to the hospice for a stay, and they reviewed his medication, he is on l0ml of diazepam twice a day, he has come home and we shall see how he goes. The school did great with the fundraising £1000, I get the total on Monday - it's fantastic news looks like we may go to Spain after all - I hope so. We had counselling at the hospice and it ready cut like a knife but it did help us to see each other's pain, it's so hard trying to be normal - what ever that is.

12th Apr 99
Well a lot has happened lately Jay has got worse he is unable to sit unaided now. He has had a gastrostomy, which is a lot better. We thought the medicine had begun to work but he has seemed very upset the last two days I hope it's nothing too serious. Tianna seems ok so far -

the next few months will tell. Jay went onto haloperidol - an antipsychotic drug but it caused fits so they took him off that and put him on Epilim, which seems to be ok. He is on 30 ml of clonazepam and still has diazepam for prn.
Life has been very mixed up and the kids are up and down I feel I have no time out ....

It is clear that life is very hard indeed with the Theobald family and we thank Fran for sharing her diary with us and we hope that Jay becomes more settled soon.

If you would like to share your story with us please contact me via email at cpm@cmartland.freeserve.co.uk or by post.

Networking

A space just to say "hello" if you want to.

Dear BDFA,

Thankyou for the newsletter.

We would like to introduce ourselves to your newsletter.

Firstly hello to everyone, My husband Richard (40) and myself (38), have two children. Our eldest son Aston is 10 years old. Our second son Louis is 8 years old and was diagnosed with having "Early Juvenile Battens Disease" in January 1996.

We have tried to carry on since then as "normal". At the moment we haven't had any contact with any other families but would like to be involved in playing a part in B.D.F.A and having contact with other families with Batten's- and them with us.

Janet & Richard Calladine
"Pumping Station Cottage", Station Road, Appledore, Kent. TN262DE
Tel:- 01233 758105

If you would like to say "hello" too, drop me a line.

I hope to get the next Newsletter to you in the Autumn / Winter sometime.
Have as good a summer as you can!

Charity Status - Postal Voting Form

The steering group committee proposes that we apply for the BDFA to become a registered charity.
To ensure that this is also the view of yourselves -the members of the association, we would like you to take part in a postal vote. Please indicate on the tear off slip whether or not you support the committee's proposal to apply for charity status.

The closing day for voting is August 24th 1999. One form per family please.

Please note, if you elect not to return your voting slips, we will interpret this as agreement with the proposal.

Please return the slips to

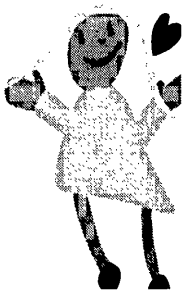
BDFA Charity Status Vote
Heather House
Heather Drive,
Tadley,
Hampshire. RG26 4QR

..... ✂
.....

please tick one box

I agree with the proposal to pursue Charity Status	☐
I do not agree with the proposal to pursue Charity Status	☐

You can supply your name, address or any other comments below if you wish



Batten Disease
Family Association

Heather House, Heather Drive, Tadley, Hampshire RG26 4QR
http://www.bdfauk.freeserve.co.uk BDFA98@hotmail.com

Patron Terry Gilliam

The Newsletter

Spring 2000 - Issue 3

Welcome to our third Newsletter. I hope everyone had a good a Christmas and Millennium as possible. Awareness of the BDFA is rapidly spreading far and wide and people are contacting us for information, advice and help, not just in the UK but further-a-field too. This year promises to be an exciting one in terms of scientific advances in Batten disease research. In September the UK is hosting NCL 2000, which will be a gathering of scientists from all over the world who are involved in Batten disease research. More on this later.

Chris Martland Newsletter Editor

Batten Disease Family Conference
Saturday 2nd October 1999

The planning and preparation for the Batten Disease Family conference, the second of the conferences organised by seeAbility and the Batten Disease Family Association (BDFA), started in earnest in July. The planning team consisted of Joan Deeley (Client Services Director), David Ireland (Appeals and PR Director), Sasha Scambler (Research Officer) and Anne Bye (PA to Client Services Director) from seeAbility, in liaison with the BDFA committee: Irena

Newcombe (Chairman), Chris Martland (Newsletter Editor), Pauline Muncey (Secretary), Sharon Burnham (Treasurer), Cliff Burnham and Andrew Henery. The seeAbility group met regularly over the next three months liasing through telephone conferences, committee meetings and e-mail with the BDFA group, and the programme, speakers, advertising, catering and a multitude of other details were sorted out along the way.

On Saturday 2nd October 1999 the Batten Disease family Association and seeAbility hosted the second Batten disease conference. The conference was aimed specifically at family members and others involved in the care of children and adults with Batten disease and the focus for the day was to provide a forum for the expansion of knowledge on Batten's disease and the sharing of experiences and information. The day long conference was the second to be held at Heather House, a purpose built residential and activity/resource centre for young adults with degenerative conditions and profound multiple disabilities, built and run by seeAbility.

The conference started at 10am with the arrival of fifty-five delegates and 8 children and young people who were warmly welcomed by David Ireland, Appeals and PR Director at seeAbility. The 8 children and young people were entertained throughout the day by Janet Dewar (Area Manager, seeAbility) and Vivian Edwards and Jenny Smith (Heather House). A programme of activities and entertainment was provided in parallel with the main conference programme, which included arts and crafts, singing sessions and the exploration of the sensory room.

Dr Irene Hofman, Head of Medical Staff at Bartimeushage in Holland, launched the main conference programme with an interesting and informative presentation with over 25 years of experience of working with children and young adults with juvenile Batten's disease. Dr Hofman provided an account of the key aspects of the care needed based on her experience of many years and containing much personal insight into the experiences of both the children and of their families. We heard about the type of care needed, the importance of sharing information with the child and the necessity of involving the parents in all aspects of the care. We also heard about some of the new drugs that are available, the importance of finding the most effective drug for each child and were given an insight into the types of hallucinations experienced by the children.

After a break for coffee, Dr Hannah Mitchison, a Lecturer in Molecular Genetics at University College London, gave the second presentation. Dr Mitchison rose to the difficult task of explaining complex genetic research procedures to a diverse audience with genetic knowledge ranging from GCSE (or less!) to MD level. We heard about the current research programme being undertaken at University College

London, the identification of the Juvenile Batten gene and the search for the mutations that cause the disease. The research into the structure of the DNA of children with Batten disease was explained and the information, although complex, was presented clearly and as succinctly as possible.

Hannah Mitchison's presentation was followed by an extended buffet lunch, giving the delegates and speakers a chance to mingle and chat before they returned to the main hall for an update on the progress and future plans of the Batten Disease Family Association. Irena Newcombe, Chairman of the BDFA, and Chris Martland, Newsletter Editor, presented the current position of the BDFA, from its inception in November of last year, through the production of the first newsletter to the printing of the first batch of leaflets explaining the BDFA and its aims. The talk covered membership and the content of the newsletter and looked at plans for the future. Suggestions were made before the debate was opened up to the floor and ideas and opinions were requested. Any ideas on the future plans and developments of the BDFA should be passed on to the committee.

The final part of the afternoon involved a panel of experts looking at a variety of social care issues. Jenny Park from Bromley Social Services gave an outline of the services available to families of children with Batten's disease and the support that should be provided by the family's social worker. Carol White followed her from Sutton Disability Benefits Centre who outlined the types and levels of benefits available, criteria for receiving benefits and gave hints on how to successfully make an application for benefits. The final speaker on the panel was Susan Green, speaking as the Development Officer for Contact a Family in Scotland, Family Support Co-ordinator and Founder of the Neimann-Pick Disease Group and parent of three children, two of whom have been diagnosed with Niemann-Pick Disease Type C. Susan gave a fascinating insight into the struggle to get appropriate education for her sons highlighting many issues faced by parents of children with Batten disease. The three presentations were followed by an opportunity for questions and debate from the floor, and with so many interesting questions and observations we ran out of time all too soon.

The day concluded with a summary session chaired by David Ireland, which included an outline of the days events and a brief note from Dr Ruth Williams from UCL about the International Conference on Neuronal Ceroid-Lipofuscinoses to be held in Oxford in September of 2000. The conference ended with a request for ideas for the future development of the BDFA, and an acknowledgement of the time and effort put in by all of the speakers taking part in the day. Tea and biscuits were served with a final chance for a chat before the day ended at 5:00pm.

At the end of the conference participants were asked to fill in an evaluation form making comments about the day. The overwhelming feeling was that the day was a success and that all of the presentations were interesting and informative. As with the last conference it was widely felt the social aspect of the day, allowing families and professionals to meet, chat and exchange ideas and experiences was in many ways the most important part of what was a successful and enjoyable day for all involved.

Sasha Scambler

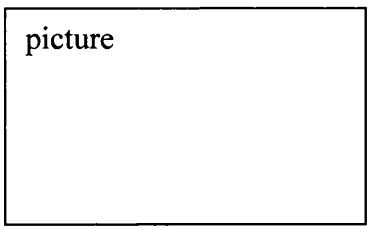
Thank you Sasha!

Everyone involved with the BDFA would like to thank Sasha Scambler for her help over the last two years. She has been a guiding influence with our group before and since its inception. She has facilitated and hosted several of our early meetings. She has kindly received and redirected our post, she has organised distribution of our leaflets and newsletters. She has also helped to organise the Batten Disease Conferences at Heather House last year. Sasha has now finished her work with seeAbility and is now working in the Public Health Department of St George's Medical School in London as a Research Fellow. She is doing research looking at the factors that cause loneliness and isolation in older people. Even so, Sasha remains in touch with the BDFA and continues to attend our meetings and provide valued help. And so we would like to congratulate and give our best wishes to Sasha in her new post.

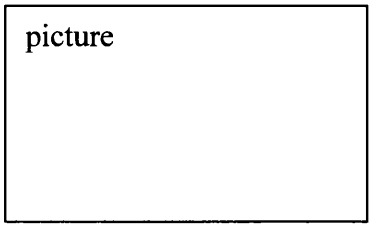
As a group we continue to be grateful for the ongoing help of seeAbility and the staff at Heather House. We are trying to stand on our own a little more now from an administrative point of view, but are still very appreciative of the support we've received.

Opening of Heather House

On Wednesday 10th November 1999, the Duchess of Gloucester officially opened Heather House at Tadley. The Duchess who is seeAbility's patron was presented with a bouquet of flowers by Helena Newcombe – a moment she will never forget. The Duchess had a tour of the building before unveiling a large commemorative brass plaque.



Helena Newcombe presents a bouquet to The Duchess



The Duchess of Gloucester unveils the plaque

She said, "I would like to say how delighted I am to visit yet another new project by seeAbility. Heather House looks to me to be a splendid result of the tremendous effort it has taken to create it.
"I congratulate all those involved in creating it through design, planning and now running this great centre. I have very much enjoyed being shown around and wish everyone all the very best for the future".
The Duchess then spent some time speaking with some of the 100 or so guests, many of whom had been directly involved in the creation and running of Heather House.

NCLRA Conference in Washington November 1999

In November Andrew Henery and Irena Newcombe attended a meeting in Washington at the National Institutes of Health (NIH), this was hosted by the Neuronal Ceroid Lipfuscinosis Research Alliance (NCLRA) who are formed of groups and parents from several countries with a common aim, to find an effective therapy which could lead to a cure for the three major forms of Battens Disease, Infantile, Late Infantile and Juvenile. These are known as CLN1, CLN2 and CLN3, respectively.
The meeting named "ACTION FOR THERAPY" consisted of a select group of scientists who have been exploring ideas towards treatment. There were representatives from the NIH and the Food and Drugs Administration (FDA), in addition there were an advisory panel of extremely talented and experienced scientists to oversee, advise and explain any aspects of the meeting and the summary produced by the NCLRA.
In these meetings scientists are encouraged to collaborate with each other and share information, this produces very good results and helps progress research more quickly.
The presentations described work being done on issues such as: CLN2 mouse models (Peter Lobel and Martin Katz), CLN3 mouse models (Martin Katz and Hanna Mitchison). Neurotrophic Factors (Jon Cooper), Stem Cell Therapy (Evan Snyder), Enzyme Replacement Therapy (William Mobley) and AAV Gene Therapy (Mark Sands).
There were also presentations from David Pearce, Jerry Faust, Rose-Mary Boustany and the FDA representative Cynthia Rask.
It is very important to stress that all this work is still very experimental and there is still a long way to go and many hurdles to be overcome before a trial of any of these still theoretical therapy can be tried, but through the efforts of these scientists and groups who form the NCLRA we are much closer.
A copy of the Summary of this meeting can be obtained Electronically

by contacting the BDFA with your E-mail address.
We also thank Ricky Bennett and Russelle Rankin who organised and put the meeting together on behalf of the NCLRA and invited us to attend.

Andrew Henery

The eighth international congress on Neuronal ceroid lipofuscinoses - NCL 2000

20-24 September 2000
Exeter College, Oxford.

This conference brings together doctors and scientists from all over the world to discuss the latest research findings. The content will probably be at a quite advanced scientific level but parents and other non-professionals are welcome to attend. There are many topics covered by many speakers including: Genetics, Functional Genomics / Cell Biology of CLN1 2 3 5 6 & 8, Clinical Aspects (Diagnostics), Therapy & Clinical Management.
One the last day, there is a session called "Parents discussion" when we will have an opportunity to ask questions and things explained.
Please see the enclosed flier / application form which gives details about costs and accommodation.

A Holiday with a Difference

Kristin Jefferson

If, like us, you struggle to find activities during holidays that are suitable for the whole family, then perhaps the Calvert Trust is the place to go. Thanks to other Batten's parents, we heard about the Calvert Trust and booked a week's holiday at the Wistlandpound site in North Devon.

Our eleven-year-old daughter Louise has Juvenile Batten Disease, and although she is still mobile, during recent months she has been ever more secure in her routine at home and at school and is not too keen on doing anything out of the ordinary. Going away can be very traumatic and stressful for her, even without any additional exciting activities. However, our younger daughter, Monica, operates at about 110% on most days, and is ready to try anything so an active holiday is just right for her.

We opted for self-catering accommodation rather than full board (but we have it on good authority that the food is really excellent). During the week we went for a walk along the cliffs,

canoed, sailed, kayaked, climbed, abseiled, braved the zip wire, rode horses (with the option of driving a pony and trap), tried our hand at orienteering and archery, and also spent many hours in the swimming pool, jacuzzi and steam room.

We all took part, including Louise, but this was thanks in large part to the skilled and sensitive staff. Monica was challenged by the activities, and had a very busy, active and sociable week. Louise started each day by saying that she did not want to take part, and went to bed each night saying what a good day she had had. The staff were gently encouraging, very supportive and always on hand to help when necessary. Emotional explosions left them totally unruffled, and so we never felt uncomfortable when things weren't going the way Louise thought they should – and didn't mind letting everybody know how she felt. The staff seemed to know when to offer help and encouragement, and when to step back (and hover!). Five minutes of their encouragement could often do more to motivate Louise than several hours of our efforts at persuasion, and her sense of achievement after each activity was wonderful. She took part in a way that we would never have believed possible and achieved things we would never have even dreamed of. We have already booked a return visit next year.

It would be difficult to say what our most treasured moment was during the week as it really was a week full of fun. However, when Monica, our younger daughter, first fearfully began to abseil down the climbing wall, it was wonderful to see her look of terror replaced by one of nonchalance as she sorted out her technique. This was followed by Louise successfully abseiling down the middle wall. When she reached the bottom she was jumping up and down and shouting about how she had managed to do what Monica had done. This was a week of many achievements for us all.

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A Day in the Life of Charlie Martland.

This piece was written by my wife Janice and outlines a typical day in our household. Charlie is 8 and has

Late Infantile Batten's disease. Maisie is 6 and is mercifully clear of the disease.

"Night blends into day. A typical night is not restful, Charlie tends to wake several times – he needs frequent suction and picking up to be cuddled. The moment Charlie shows signs of waking I move him, a nudge is enough, and I hope desperately to see the edges of a smile - a good indicator of how he is - how the day will unfold, no smiles and sometimes worse than that; wretched coughing, reflux and upset, heralds a bad day. My heart sinks and I do my best to console him. In between all of this Maisie wakes and needs love and cuddles and attention. She gets into bed with Charlie and myself, and we have mutual loving and holding. If she doesn't get this attention she will be very grumpy! Alongside all of this, Chris tries to co-ordinate breakfast, cups of coffee and thoughts about school and work - essentials that have to carry on despite our personal desolation generally, and despite any specific crises that may have unfolded and developed overnight or whenever.

Working against the clock, we force our aching, tired bodies into action and get washed and dressed with minimal attention for ourselves. We jolt brains into gear and try and think what day is it? What's happening today?

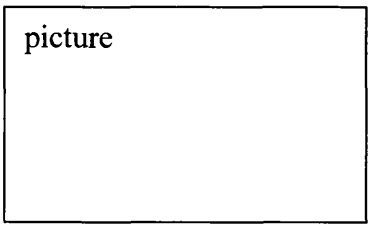
Charlie's needs are uppermost - fluids, nutrition, medicines, bathing, dressing, cuddling - all of which take up most of my time all day. Chris now baths Charlie after I strained my back and shoulder doing so - he's a heavy boy! But we try and take it in turns to do the lifting and carrying. Charlie loves his bath - we get it as hot as possible for him, so that he can soak and relax all his muscles and joints - he feels so stiff and uncomfortable first thing - but after his bath he seems better.

Quite often when Maisie has gone to school and Chris to work, Charlie will sleep for a couple of hours - this gives me time to do essential jobs, clean up the house, hang washing out etc. But I often just sit down next to Charlie in his chair, put some favourite music on, look at him, watch him breathing, and think about the whys and what fors, and maybe shed some tears, and do some grieving.

This kind of routine is only new to the last few months. The previous two years have shown us different extremes each day or night throwing up new situations, new crises, and new problems on a daily and sometimes hourly basis. Looking back, you wonder how you get through the difficult times but you just do, and each "remission", or each stable period although grasped and clutched with much gratefulness and thankfulness, only heralds another spiral downwards into the blackness of this dreadful all consuming disease. And so we feel a little like sitting ducks, - just waiting.

But the good bits give us time to recharge our batteries - and prepare. And that's what we do.

Charlie wakes and the day carries on. I plead for good weather so I can get him out for a walk in his wheelchair- he loves being out and about, and so that's what I try and do. Complete with suction machine, fluids, sometimes medicines and the mobile phone just in case.



Charlie age 4, just after the diagnosis

School proves too difficult now - transport is not possible, and he needs constant care. I feel it is my duty to provide that care - I know school can manage to give it, with extra funding for nursing care but I brought him into this world - he is mine to look after, to love and to ensure all his needs are met. To ensure his passage through this hostile life is as good as it possibly can be. And to be there when the flame dies and for me to carry him out.
Maisie comes home from school and reality starts up again - "normal" life as we occasionally glimpse it - has to kick-start again - for her sake. We make tea, we play games, read books, give her praise, tell her off etc etc.

"Normal" life takes a dive when Charlie is ill - but Maisie adapts well. We involve her, we are truthful with her, and we try desperately to protect her from the harmful emotional / psychological effects of this hell. Sometimes this is very difficult.

Without help from our local hospice (Martin House), Crossroads and friends, we might not cope as well. They provide us with a lifeline and some glimpses of sanity.

Then it's bedtime for Maisie, more cuddles, and story reading, laughing and loving. Charlie drifts off later if we are lucky. When he sleeps, I sleep with only one eye closed. I hear every sound, every change in his breathing pattern, and know instinctively when something is wrong. Day blends into night. The cycle carries on."

Douglas House – *A Respice for Young Adults*

Sister Frances Dominica; the founder of Helen House and now Douglas House in Oxford is responsible for the core philosophy behind the Children's Hospice development throughout the country. Her selfless approach and devotion to children, whose lives are coming to an end, and their families, is remarkable. Her unconditional concern and love for fellow human beings inspires and humbles us all.

In the 17 years since the first children's hospice, Helen House, opened many things have changed for children with life-limiting illnesses. Not least among these is the option of the introduction of gastrostomies for children whose swallowing reflex is impaired. Adequate nourishment often means a longer life span than would otherwise have been likely. Another example is children with cystic fibrosis for whom intensive physiotherapy and a fat restricted diet means life may be extended to early middle age.

Over the years we have been increasingly concerned about the inappropriateness of a children's hospice for young adults. A six-foot, fourteen stone 21 year old young man in the hyper-active phase of one of the mucopolysaccharidoses needs a different environment for respite care from that which we can provide.

An additional factor in our thinking has been that most children's hospices do not take new referrals over the age of 16. We are

increasingly aware that some young people the need for respite care or terminal care has not become apparent by the age of 16 and that when they do reach the stage when they need such support there is little or nothing on offer.

We do not envisage a high proportion of young people with cancer using Douglas House, any more than is the case with children's hospices. Rather it may be used by young adults with the genetic conditions with which we are already familiar – MPS, Batten's, Duchenne muscular dystrophy, cystic fibrosis, congenital heart disorders, for example – as well as such illnesses as motor neurone disease and HIV related illnesses.

Douglas House is named after a young man who had juvenile Batten disease and became a great friend during his 80 or so stays with us at Helen House from the age of 15 to when he sadly died aged 24. Douglas had a sister; Penelope who also had the illness. It will be for young people between the ages of 16 and 40 and will offer outreach services, day care, respite care (with the provision of 6 or 7 beds) terminal care and bereavement support. We will respect young people's desire to be as independent as possible and will be sensitive to the fact that while parents may still have a practical as well as emotional involvement partners may have taken on a vital role. We plan to provide recreational facilities and up-to-date IT equipment to enable study and ease of communication.

Why is it called a "respice"?

Seventeen years ago David Baum brought a visiting neo-natologist from California to Helen House. He suggested we call Helen House a respice on the basis that where there isn't an appropriate word it is good to create one! Hospice, with its connotations of terminal cancer, has not always stood us in good stead. So now, all these years on, we have the courage to take Bill Silverman's advice. Douglas House will be a respice. It was David who, many years ago, told me the Starfish Story, which I now see as epitomising his own philosophy of life, a life cut tragically short when he died on 5 September 1999. He was President of the Royal College of Paediatrics and Child Health and was on a college sponsored cycle ride to raise funds for the children of Kosovo and Gaza when he had a massive coronary thrombosis. He was an excited and enthusiastic supporter of the Douglas House project and in affection and gratitude we have adopted the Starfish as our logo.

> *As the old man walked the beach at dawn, he noticed a young man ahead of him picking up starfish and slinging them into the sea.*
> *Finally catching up with the youth, he asked him why he was doing this. The answer was that the stranded starfish would die if left until the morning sun. "But the beach goes on for miles and there are millions of starfish" countered the other.*
> *"How can your effort make any difference?"*
> *The young man looked at the starfish in his hand and then threw it to safety in the waves.*
> *"It makes a difference to this one," he said.*

There is something very special in each and every one of us. We have all been gifted with the ability to make a difference. And if we can become aware of that gift, we gain through the strength of our vision the power to shape the future for young people with a terminal illness.

Together we can make a difference.

Sister Frances Dominica
Founder and Director of Helen House and Douglas House

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Lucy's Poem

Suddenly I'm in a place where everyone has a smiley face, and I can see them smile,
I don't know how I got here or even why I came,
But it does seem very, very nice, and if you don't mind, I think I'll stay.

Darling mum and dearest dad, I know that you are very sad,
But I'll always be your daughter, your precious eldest child,
A sister, niece, a cousin, a dearly loved grandchild.

And lots of little children are playing merrily,
They're waving, smiling, calling me to join in all the fun,
And my arms can wave, my legs can walk, and I can laugh and I can talk,

If only you could see me now you'd not believe your eyes.
Your lovely child can skip and sing! I can do just anything,
One day you'll come and join me in this happy sunny world,

So until then, my dearest mum and darling dad, please be glad for me,
For I can shout and jump and run, at last I'm free,
I'm free!!!!!!!!!!!!!!!

This was sent in by Martha and Jonathan Horsford from Tunbridge Wells in Kent, the poem is about their daughter Lucy May who died aged 5 on 26th January 2000 from Infantile Batten Disease. Martha said that the poem was sent to them after Lucy died and it helped them a great deal, she has sent it in with the hope that it may help other parents.

Membership Subscriptions

We've now been up and running for about twelve months which sadly means we need to ask for everyone's annual subscription. This remains at £10 and remains voluntary. We hope you understand that at present we do need to have this system to keep the administration ticking over. Please find a membership form enclosed, this can be used to renew your subscription or change any details so we can keep our files up to date. If you haven't joined already then - jump aboard!

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