Stories in Isolation

The impact of gene discovery on families and professionals

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Submitted to Cardiff University in fulfilment of the requirements for the degree of Doctor of Philosophy

September 2009

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Acknowledgements

Thanks are due firstly to the families for their time, energy and generosity in giving very personal interviews, and the scientists at the Institute who shared personal and professional memories of their endeavour.

My own family have been unstintingly generous in their support. Special thanks to Katie for proving that star charts work for mature students as well as toddlers, Ciara for pep talks at crucial times, and Julian for astute comments and mastering beans on toast when time was short. My dad and sisters Orna, Hilda and Anne have, as always, been a source of comfort and diversion when it was needed. Mum, though absent, is constant.

Thanks to my supervisors; Paul Atkinson at the Cardiff School of Social Science (SOCSI) whose insights guided not just the research but me as a researcher; and Angus Clarke at the Institute of Medical Genetics and the ESRC Centre for Economic and Social Aspects of Genomics (Cesagen), for his optimism and support. Mel Evans at Cesagen averted several technology related crises and the library staff, especially at the Bute, were always helpful. Thanks to Jamie Lewis, Sara Delamont, Maggie Gregory and Richard Taulke-Johnson for perceptive advice, and to Katie Featherstone and Hannah Lister for ensuring that the last few steps were taken. Liz Renton at the SOCSI Post-Graduate office has been patient and kind, no matter what the enquiry.

It would not have been the same experience without the friendship and support of Rebecca Dimond, Amy Lloyd and Jackie Needs. To my old friends, especially Lee-Anne, Caroline, Carol, Maria, Lorna, Pauline and Jo, thanks for not giving up on me when I was preoccupied, and to Sian for neighbourly help.

Thanks to Peter Harper whose help was always given kindly and to Lindsay Prior who gave me an unexpected opportunity.

This research was supported by ESRC/MRC studentship PTA-037-2005-00015

Summary

This thesis examines the impact of the 1992 myotonic dystrophy gene discovery on families, clinicians, and a team of scientists who played a key role in the successful international research collaboration. The gene isolation resulted in a diagnostic test but not, as yet, treatment or cure. The scientific team, now dispersed, and families attending the myotonic dystrophy clinic were interviewed, and the myotonic dystrophy medical record archive was examined. Reflexive practice enabled the research strategy to adapt to emergent themes. A broad repertoire of qualitative methods was used to explore the data from these varied sources.

Documentary traces in the archive captured research and service trajectories, from the grounding of scientific success in relationships between home, clinic and laboratory, to the contemporary management of myotonic dystrophy where bureaucracy and technology are visible but clinical expertise predominates.

Through vivid recollections and use of narrative devices the scientists reconstructed a unique era in clinical genetic research. An emotional register, privileging relationships and the grounding of scientific advance in everyday laboratory work, distinguished their accounts. This language revealed subtle differences between narratives, where there was universal recognition of the importance of the discovery for a scientific career, but ambivalence regarding its personal meaning for some key actors.

For families, gene discovery represented hope for future generations while personal meaning was located in the maintenance of valued roles of everyday life. The accounts narrated the challenges of adapting to an uncertain prognosis despite definitive diagnosis. Vocabularies of strength were at variance with physical weakness highlighting the significance of narrative analysis as both method and representation of meaning.

Analysis of gene discovery revealed complex interpretations of meaning for the scientists, multiple representations of myotonic dystrophy across the data sources, and the gene test, rather than gene isolation, as a key turning point for families.

Contents

Introduction and Structure

The discovery of the gene for myotonic dystrophy (DM) by an international research collaboration in 1992 represented a major scientific advance. A small team of scientists, based at a local Institute of medical genetics and led by a clinical geneticist, made a significant contribution to DM gene discovery through a combination of clinical and scientific expertise and cooperation with an extensive network of families with DM.

A tendency towards earlier onset and increased severity of DM in successive generations had been observed clinically but the biological basis of this phenomenon, known as anticipation, had been fiercely contested in the decades before the "era of human clinical genetics" (Worton 2001, p. 819). The isolation of the DM gene on chromosome 19 resulted in the identification of the molecular mechanism for anticipation. This unstable triplet repeat mechanism in the myotonic dystrophy gene is not yet fully understood but gene isolation resulted in a diagnostic gene test for DM almost immediately.

However details of the everyday work that went into the DM gene discovery and the everyday reality of living with DM have largely remained undocumented. "Stories in Isolation" refers to DM gene isolation and also to the way scientists' and family accounts extend scientific and medical descriptions of DM.

This thesis presents multiple accounts and trajectories of DM, in particular highlighting the research design necessary to interrogate a variety of data sources. There is a consistently methodological strand to the thesis, which is structured around seven chapters.

Chapter one, "Stories in Isolation" introduces the DM gene discovery and the background to the research question through a description of the event, an outline of the resources used in the project, an overview of DM in relation to scientific research and medical classification, and the current focus of DM scientific research.

Chapter two, "Bodies of Work" places the research question in the context of previous work through engaging with relevant literature and analysing potential key areas of enquiry. The sections within the chapter reflect the necessity to engage with a broad range of relevant literature due to the interpretive possibilities offered by three different sources of data. The initial sections refer to sociological research at the time of DM gene discovery, the significance of discourse to the sociology of science, the importance of practices and processes in offering insight to scientific culture, and the engagement of families with research. Research relating to disability, chronic illness, narrative, and the sociology of the body is then explored. This addresses the importance of remaining sensitive to the context of the family interviews through accounts given by the individuals themselves rather than imposing a pre-defined narrative structure. The importance of the DM archive is highlighted through a review of key authors in the field of documentary analysis and the use of medical records. The clinic as the site of interaction between the family and the clinical geneticist is explored through analysis of literature focusing on areas of activity such as classification of DM, the management of uncertainty and the impact of technology. Inherited conditions from the family perspective are analysed through theories of kinship and risk.

Chapter three, "Characterising DM" explores the conduct of the research and has four main sections. The first introduces the research background and resources. The second describes development of the conceptual framework and research design. This recognises the many possibilities for design and interpretation of the research question. Through reference to the work of others decisions were made about methods and there was the first real ownership of the project. The third section reflects on the process of putting this framework into action in the real world, with all its contingencies and unforeseen circumstances. The fourth section addresses analytic choices for the three data resources.

Chapters four, five and six report the analysis.

Chapter four, "Molecular Detectives" explores interviews with six members of the original DM gene discovery team in relation to themes of scientific culture, turning points, dynamics, discourse, boundaries, personal meaning and the legacy of discovery. Through narrative analysis the perspectives of the key actors emerge.

Chapter five "Bound Together" focuses on the trajectories of research and service documented in the DM medical records. Unprecedented access to the archive, which functioned as an artefact of the past and a core component of contemporary clinical genetic practice, highlighted the incremental and mutually dependent nature of DM research and service over three decades. The analysis comprises two sections. The first utilised an analytic framework investigating the different kinds of activities visible in the notes to examine three major themes. The work of the clinic, the experience of families, and the representation of DM were analysed through exploration of the relationships underpinning the documented work, the management of risk, and the classification of DM. Analysis highlighted the significance of metaphor and offered an opportunity to examine it in the context of everyday work. The second section examines one key set of notes through a narrative analysis and explores how essential research cooperation with families was accomplished through devices such as rhetoric.

Chapter six, "Gripping Stories" examines the interview accounts of twenty people with a diagnosis of DM. Themes of personal biography, the challenge of retaining agency, personal meaning as expressed through narrative, and the impact of physical ability on performance in social situations are explored.

Narrative analysis of interviews with the local team of scientists and with members of twenty families with DM, revealed complex and emotional responses to everyday work and everyday life with an unpredictable and incurable hereditary condition. Scientists told stories that were not reflected in official scientific discourse. Families constructed pragmatic narratives of hope for research in the future but narrated personal meaning through accounts of everyday life. Analysis of the interviews

highlights how the published scientific account, and textbook medical definition are the dominant, but not the singular story of DM.

Appendix four contains family trees outlining the pseudonyms and ages of the family members interviewed.

Chapter seven, "Everyday lives, Genetic tomorrows" examines the analytic findings through a discussion of three main themes followed by a short final summary. The themes relate to the use of narrative to examine the implications of DM in the context of everyday life and everyday work, the unusual focus on the emotional aspect of groundbreaking scientific work apparent in the scientific interviews, and the necessity of researching scientific advance from the perspectives of those on whom it has most impact.

Multiple representations of the events of gene isolation and of DM were elicited through exploration of the impact of gene discovery. These different interpretations illustrate "those life experiences (epiphanies) that radically alter and shape the meanings persons give to themselves and their life projects" (Denzin 1994, p. 510). The transformative narratives of the family members, the multi-faceted narratives of the scientists and the rich material of the DM archive highlight the complexity of dynamics influencing both research and management of a condition that continues to be characterised by uncertainty even after gene discovery.

Chapter One

Stories in Isolation

Introduction

In 2008, sixteen years after the event, a scientist recollected her shaking hands as she looked at the positive results of a laboratory experiment in the final stage of the race to identify the gene for myotonic dystrophy. Isolation of the DM gene by an international collaboration of scientists in 1992 represented one of the defining stories of the narrative of the condition itself, and of the phase of disease gene identification that Conrad and Gabe (1999, p. 1) referred to as "the dawn of a genetic age". The scientific account of the isolation of the gene for myotonic dystrophy on chromosome 19 was published in prestigious scientific journals such as Cell and Nature (Brook et al. 1992, Harley et al. 1992). The close submission and publication dates tell the story of the highly competitive environment surrounding public disclosure of the event.

> Myotonic dystrophy is the commonest adult form of muscular dystrophy, with an estimated incidence of 1 per 7,500, although this is likely to be an underestimate because of the difficulty in detecting minimally affected individuals. It is a multisystem autosomal dominant disorder of unknown biochemical basis. No case of new mutation has been proven. We have isolated a human genomic clone that detects novel restriction fragments specific to individuals with myotonic dystrophy. A two-allele EcoR1 polymorphism is seen in normal individuals, but in most affected individuals one of the normal alleles is replaced by a larger fragment, which varies in length both between unrelated affected individuals and within families. The unstable nature of this region may explain the characteristic variation in severity and age at onset of the disease. A second polymorphism at this locus is in almost complete linkage disequilibrium with myotonic dystrophy, strongly supporting our earlier results which indicated that most cases are descended from one individual mutation.
>
> Harley et al. 1992

Widespread media and scientific interest accompanied publication of the definitive account of discovery as the underlying biological mechanism explaining the highly variable phenotype and complex inheritance pattern was revealed. Images of scientists in white coats looking down microscopes encapsulated the public image of the molecular detective, while accompanying stories of families with DM lent human interest. Corridors which had in the preceding months been lit day and night as scientists worked around the clock were briefly crowded with the unfamiliar presence of TV crews and journalists, before the spotlight moved on to the next story and the next phase of science. Newspaper headlines such as "Dystrophy Gene Nailed" signified the discovery as one of the popular media stories of the emergent

era of clinical genetics (St Paul's Pioneer Press 1992). The headline captured the notion of a disease- causing gene as a deviant phenomenon with independent agency that could be overcome through scientific pursuit (Myers 1990).

Isolation is the term often used to describe the identification of a gene. The 1992 DM gene discovery was part of a unique phase in clinical genetics where the molecular basis for hereditary conditions caused by single genes was being established. This did not happen in isolation from families with DM whose contribution was crucial to the success of the local team. This thesis reports on this key juncture in the trajectory of DM through the memories and accounts of the local team of scientists and clinicians, families attending the local DM clinic and the histories documented in the DM medical record archive.

The archive is located at the local Institute, where the family members interviewed attend a specialist DM clinic. The team of scientists, led by a clinical geneticist, had been based at the Institute during the time of gene discovery but the team subsequently dispersed and were in various geographical locations when interviewed. The clinical geneticist who set up the Institute had overall leadership of the local gene discovery team. His contribution and influence was succinctly articulated by one of the scientists who referred to him as "Mr Myotonic". He has been retired for several years but contributes actively to the field of human clinical genetics through his work on the history of this now established speciality. Key actors from the scientific team and family members interviewed have been anonymised and given pseudonyms. The geneticist is referred to as Prof, the senior scientist as Andrew, the two post-doctoral scientists as Pauline and Tom, the laboratory technician as Kay and the PhD student as Bill.

Access to a developing specialist centre for muscle disease, combined with relationships with families built up through research, combined to establish an approach characterised by in-depth knowledge of the everyday manifestations and natural history of DM. The DM gene discovery reinforced Prof's reputation as an internationally recognised expert in DM and the Institute as a centre of both scientific and clinical excellence.

The sociological significance of DM gene discovery was explored in the aftermath of isolation and publication. The local team were the subject of ethnographic fieldwork and semi-structured interviews. Subsequent publications related to the processes of competition and collaboration and the influence of personal dynamics on the field of scientific inquiry (Batchelor et al. 1996; Atkinson et al. 1997; Atkinson et al. 1998). This work extended the pioneering work of Gilbert and Mulkay (1984) on scientific discourse and asserted "we know too little about the processes of transmission of innovative knowledge from the benches of laboratory scientists to the bedsides of patients and the consulting rooms of medical practitioners" (Batchelor et al. 1996, p. 248). The "fluid" (Atkinson et al. 1998, p. 279) nature of allegiances and the validation of discovery within the scientific community offered valuable insight into the complexity of the everyday work that underpinned a major scientific advance.

The contemporary accounts given by the scientific team highlight emotions varying from pride to ambivalence, contrasting with the accounts of gene isolation which were written in the accepted format for scientific publications, removing agency and subjectivity. In their retrospective accounts of DM gene discovery the scientists reconstruct the significance of relationships, problems and dynamics in a way that the definitive scientific account did not. In narrating the real life contingencies of scientific discovery, the scientists observe that these aspects were not usually reflected on within official scientific discourse. The narratives of gene isolation illustrate how the process underpinning scientific accomplishment remains isolated from the discourse documenting the accomplishment itself.

The use of narrative analysis was based on the ubiquity of storytelling as a way of constructing and conveying meaning (Polkinghorne 1988; Bruner1990; Coffey and Atkinson 1996; Ochs 1997; Denzin 2000; Elliott 2005; Riessman 2008). Implicit in this was the recognition of possible alternative accounts or additions to the official scientific account, and to forms of discourse such as the illness narrative. Analysis focused on language as a way of asserting individual meaning in describing a process of major scientific progress, and the experience of living with an inherited, incurable condition that already has standardised descriptions in medical and scientific literature.

The contemporary landscape of clinical genetic research is concerned with the complexity of genetic and gene-environment interaction rather than with identification of single genes. The DM gene isolation resulted in a diagnostic gene test almost immediately, representing a successful interdisciplinary translation. However there is currently no cure for DM, and the initial hype and hope that gene identification would lead rapidly to treatment has been tempered by acknowledgement that the process will be lengthy. Watson's assertion (1989, cited in Muller-Hill 2002, p. 928) that "We used to think our fate was in the stars. Now we know that in a large part it's in our genes" remains as simplistic as Conrad and Gabe's heralding of a new age (1999). Greater knowledge about the natural history of DM has resulted in significant improvements to the medical management of aspects of the condition such as cardiovascular and respiratory care. However, the uncertainty relating to diagnosis that characterised clinical work prior to DM gene discovery has been replaced by uncertainty about prognosis.

Beginnings

The DM work began in the early 1970's with the arrival of Prof as a young doctor to set up a department of medical genetics. As part of establishing this new approach to medicine Prof began to give talks on his previous work with the renowned geneticist Victor Mc Kusick in the US, in addition to taking referrals for genetic counselling. In one of his first talks he spoke about his interest in DM and showed a photograph of a person with the condition. A paediatric neurologist approached him afterwards to discuss a child, Ben, whom she thought might have DM. This referral, which also resulted in the referral of Ben's mother Alys for genetic counselling for her second pregnancy, signified the beginning of the DM research and service in the department. Having thought that there would be little opportunity to continue with DM research Prof now embarked on a study investigating myotonic dystrophy in childhood and the first DM medical record was put together.

The scientific team who became part of the successful collaboration to find the DM gene were based in laboratories that formed an integral part of a developing centre for clinical genetics.

It was quite a small outfit and I don't know how many people. Probably half a dozen in the department. We were all crammed into a little lab, I remember that, and we had to get a lot of technology and stuff off the ground.

Andrew

Transient presences

Trainee doctors played a significant role in crossing the boundaries of the laboratory, family home and clinic. Their contributions to the medical records marked a unique time of research progress, where resources associated with the developing profile of expertise in DM enabled the deployment of personnel to research and clinical management of the condition. They are referred to as junior doctors in this thesis, reflecting the hierarchical nature of the medical career. These doctors were at varying stages of seniority and their funding was not from a single predictable source but reflected the nature of resourcing research work and its dependence on applications to a variety of funding sources. Their transient involvement emerged from the reconstruction of the scientific accounts and their inscriptions in the medical records.

They marked an era in DM research at the Institute that has ended and the presence of these junior doctors gradually disappeared in the years after gene discovery. The medical records document the profile of the DM clinic from its origins in the joint enterprises of research and service, and populated by professionals reflecting these dynamics, to its current status as a specialist clinic run by two senior geneticists, with no trainee doctors or ancillary staff other than those fulfilling vital administrative roles.

Archiving the past and illuminating the present

I enclose blood samples...kept in a fridge overnight...I hope they're in good condition.

Handwritten letter from family member 1975

The DM archive documents a changing picture of DM, reflecting increasing incorporation of knowledge into clinical practice. The records mark key turning points such as the translation of the gene discovery into a diagnostic gene test. They also testify to the ongoing challenges of accessing resources to facilitate the maintenance of everyday life for people with DM. The contribution of artefacts such as photographs, letters and family histories vividly illustrates a time when families

engaged with DM research in their homes, and presented an alternative picture to the traditional concept of laboratory based research.

The records contain letters from families to doctors and doctors to families, referrals and consultation details from doctors in one speciality to another, family trees and clinic record sheets, all of which have their own norms of composition. They are filed together providing a place where the everyday work done by scientists and geneticists intersects with details of the everyday effects of DM on families. The process of discovery of the DM gene over several decades and the subsequent impact has been documented. Now, through the traces left of scribbled directions and telephone numbers, handwritten letters with invitations to supper, and letters describing how blood samples had been kept in the family fridge overnight, it is possible to glimpse a unique and largely vanished era in medical genetic research. In this intersection of the everyday work of the geneticist and the everyday life of the family the cooperative nature of the research work becomes visible. With the advent of gene isolation and a definitive DM diagnostic test the contemporary challenges of practising and experiencing the familial implications of a genetic condition, whilst acknowledging the needs and rights of the individual, could be traced.

The DM archive documents the gradual accumulation of knowledge about the course of DM achieved through interaction with families, and the explication of DM as it is experienced in everyday life. The incremental application of knowledge to practice, and the way the implications of scientific advance were apparent in issues of ethical and social importance, occurred alongside the progress towards gene discovery. The medical records indicate how these challenges were responded to before there was awareness of them beyond the immediate medical context, and how these decisions shaped current practice in areas such as predictive and childhood testing, and the relationship of personal genetic information to insurance and employment.

Interpretations of progress

The interview accounts of family members related to the gene test resulting from DM gene discovery, rather than to significant memories of gene discovery as scientific progress. Gene discovery was part of a narrative of pragmatic hope rather than a memorable event. The gene test made possible by isolation of the DM gene

represented certainty and definition in the course of an otherwise variable and unpredictable condition.

> *It wasn't in their notes that I had it [DM], because they hadn't taken a blood test.*
>
> *Michael*

The way Michael, a grandfather, spoke of how his father had been affected, and his worries for his daughter and granddaughter, gave insight into the stress of living with an inherited condition. His interpretation of the definitive status of the genetic test, despite clinical confirmation of DM two decades before, portrayed how the official narrative of DM is translated through family experience and understanding. This personal interpretation of the mystery surrounding diagnosis was relived in the way his daughter as a child had asked to be seen at the DM clinic in case her knee pain was associated with her father's muscle weakness, yet declined definitive testing as an adult. Confirmation of her unaffected status came as a result of an emergency referral by a midwife during her second pregnancy, rather than as a response to repeated invitations to have genetic counselling from the DM clinic. In this way the story of DM through the generations and its familial understanding can be traced, and the intersection between the clinic and the home explored as the site of interpretation. Michael's reference to the "notes" reflected how the medical records are referred to and used in their natural setting as instruments of everyday clinical genetic practice.

Family members referred to DM as a condition. There was resistance to its definition as a disease. The isolation of living with a poorly understood condition was described. Paradoxically, narratives of accomplishing meaningful lives also represent stories of isolation in the face of a condition sometimes described by negative reference to personality and character in addition to physical deterioration. The variability of the phenotype, resulting in varying manifestations of the condition even within the same family, further exacerbates uncertainty about the future. There was continual negotiation between the need for a diagnosis in order to access resources, and the ambivalence associated with reduced expectations of capabilities by others and possible stigma through invoking the DM diagnostic label. Uncertainty about the future was conveyed through loss of agency and a relinquishing or reinterpretation of anticipated roles for the future. The achievement of meaning though incorporating

the uncertainty of DM into personal biography was active, challenging and ongoing, adding another dimension to the gradual physical deterioration and apathy characterised in the medical classification.

Family members indicated their awareness of the manifestations of the condition and how this related to the maintenance of relationships and self- presentation. They narrated awareness of the reduced capacity for social interaction due to the involvement of facial muscles and speech. Reference to characteristic apathy in medical articles was represented in the family interviews by insightful narrating of what it was like "to lose your get up and go" or by a partner describing the clinical observation of excessive daytime sleepiness as being like "living with the dead". Rather than focusing on gene discovery family members used the interview opportunity to narrate DM from the perspective of coping with it in everyday life. These powerful representations provide a different set of accounts and histories to the medical classification of DM and the scientific discovery of the DM gene as their meaning was explored through narratives of family experience.

Partners and spouses spoke of the challenges of being in a relationship where there were caring responsibilities in addition to diagnostic and reproductive implications for the extended family. There were feelings of isolation resulting from the focus on the person with DM, in addition to the loneliness of living with a person whose condition resulted in excessive tiredness and weakness. Reluctance to disclose symptoms to other family members due to a wish to protect them and to maintain independence and autonomy over valued roles led to feelings of isolation within relationships in the early stages of the condition. In other situations an inability by the unaffected partner to come to terms with DM diagnosis led to marital breakdown. The narratives varied in context and content, reflecting the variability of the condition and the situations in which it was being expressed, but were consistent in their linguistic assertiveness, and thematic concerns of loss of agency and uncertainty about the future. Memories of previous abilities and the relinquishing of valued roles within the family and community were evoked through stories of past activities. The reconfiguration of expectations to accommodate the trajectory of DM alongside the trajectory of personal biography gives personal meaning to the DM diagnostic label.

The perspectives of the scientific team were distinguished by detailed recall of the events leading up to gene discovery and of the relationships that were integral to the process of discovery. There were important differences too, reflecting hierarchy and gender, but the gene discovery as a significant event on both a career and personal biography was universal. Recognition of the personal meaning of gene discovery produced more divergent accounts, ranging from statements such as "I haven't done anything better since" to accounts contrasting it with other, more motivating, work.

Narrative analysis was used to interpret the legacy of gene discovery and to explore the linguistic devices utilised to narrate the trajectory of discovery and its subsequent impact. The scientists spoke of discomfort evaluating events of the past, attributing this to a lack of familiarity reflecting on past events in a profession where the accomplishment of scientific work is focused on continual progression from one project to the next. Their accounts were marked by the emphasis placed on dynamics and friendships and the legacy of gene discovery on their subsequent careers.

Prof's account focused on gene discovery as part of a perspective encompassing a career in the research and clinical management of DM. In describing his involvement as "a great privilege" he emphasised how close involvement with families with DM over decades had informed his perspective on the effects of the condition on everyday life. The importance of issues such as competition and collaboration within the academic scientific community were recounted with an overview of how they were accomplished and lessons learned. The significance of gene discovery was narrated beyond the laboratory to its impact on families and the clinic. Gene discovery as a turning point reinforced the perspective of ongoing work where gene isolation was an essential step towards providing further knowledge and better management for families. The engagement of scientific progress with clinical situations and the ethical issues that arose out of advances characterised a narrative of reflective evaluation.

A factor common to the scientific and family narratives, and to the documenting of DM over several decades, was the grounding of meaning in everyday work and relationships. The scientific team attributed meaning to recognised success, and to

the experiences that had led to success, whilst family members expressed meaning in the continuation of lives rooted in valued roles of everyday life.

The department set up by Prof in the 1970's is today a large Institute with an international reputation and responsibility for delivering a regional clinical genetics service. Physically it has grown and moved from a small corridor within the main hospital to a large purpose built building that it has outgrown, necessitating the housing of some NHS staff in other buildings on the hospital campus. The physical proximity of the scientific and clinical teams working on genetic disease embodies the original philosophy of combining resources and knowledge to gain an overall perspective on genotype, phenotype and implications for families with genetic conditions such as DM.

The story of myotonic dystrophy

2009 marks "100 years of myotonic dystrophy" with events signifying progress in classifying, researching and managing "a syndrome that to this day plays an important role in neurologic practice" (Steinberg and Wagner 2008, p. 961). Defining DM has been contentious since its first medical classification after Steinert in 1909 "...on the basis of precise observation, subtle neurologic examination, and great clinical experience" (Steinberg and Wagner 2008, p. 961). The complexity of the inheritance pattern, called anticipation, whereby DM was clinically observed to become worse as it passed through generations of a family was contested through the twentieth century and was finally resolved through gene discovery in 1992.

Whilst the centenary of the classification of DM by Steinert in 1909 has resulted in an international anniversary conference, the juncture of the DM diagnosis with individual biography resulted in personal anniversaries of enduring significance, recounted at length and in detail during family interviews. Memories of diagnosis replayed through accounts of the search for information about an uncertain future.

In the process of investigating the impact of DM gene discovery through medical records, and scientific and family interviews, it became apparent that there were many accounts of DM to be found. The context surrounding the gradual evolution of

DM emerged, from its recognition as a distinct classification, through the process of refining this classification, its role in the social as well as medical ideology of the first half of the twentieth century, to its current representation as an exemplar of a inherited condition with a novel molecular mechanism. The uncertainty surrounding DM diagnosis due to the variable phenotype has been resolved due to the DNA blood test developed after gene discovery. However the locus of uncertainty has now moved to prognosis.

> *They say can't give you like a time scale see...Seven years or ten years and they go no, well no. And I need to know...And I really need to know...But they, they can't –*
>
> *Louisa aged 39 DM family interview*

Exploration of the impact of advances in DM knowledge on the experiences of families and professionals reveals a rich picture of the contingencies and challenges of living with, researching, and managing uncertainty. The complex accounts of the discovery of the DM gene, the experience of the condition for families and the representations of DM in the medical record reflect the insufficiency of a single narrative to convey the multiple meanings of this inherited, incurable and degenerative muscle disease. In re-telling the stories in gene isolation the emerging narrative of DM over time becomes apparent

Medical representation of DM

DM is the commonest muscular dystrophy of adult life. It is highly variable in its manifestation, severity and age of onset. Congenital DM, childhood onset DM and adult onset or classical DM are all classified under the umbrella term DM. There is multi-system involvement of smooth muscle, the heart, brain, endocrine system, eyes, skeletal system and the skin. There may be a "typical" countenance due to muscular weakness in the face causing the jaw to hang open and the eyelids to droop. Male pattern baldness is common. Cardiac complications, notably arrhythmias, are the principal cause of sudden death in DM, whilst the overall main cause of death is cardio-respiratory complications (Harper 2001, 2002).

The medical classification of DM represents the multi-systemic effects of the condition and this was reflected in the medical records by the documenting of "typical" presentations. Although the condition is noted for its variability there are

characteristics that define it clinically such as myotonia, manifesting as a difficulty releasing handgrip. However, since the advent of the gene test there are also instances of pre-symptomatic testing where clinical examination did not reveal signs or symptoms.

Myotonic dystrophy has been defined in different ways depending on the medical and social context of the time. The initial process of classification was followed by phases of concentrated research into its characteristics, in accordance with the clinical expertise and technologies available. This included focus on the emerging evidence for more than one type of DM. The variability of the phenotype and the unusual inheritance pattern perplexed and polarised the geneticists of the twentieth century until the gene for DM was discovered.

Classification

The nosography of DM reflects changing traditions and practice over time. DM was also known as Steinert-Curschmann disease, reflecting the significance of the contributions of Steinert and his mentor at Leipzig University hospital. Steinert died aged 36 but his work was recognised posthumously and promoted by Curschmann. The classification and naming of DM made it visible, and through the influence of a well-recognised doctor information and awareness about it was disseminated. In the medical literature of the period in England DM was also associated with names such as Batten and Gibb, and in Russia with Rossolimo.

Batten and Gibb's 1909 paper drew upon their clinical observations and others in the literature to state "we think the type worthy of recognition" (p. 187). This "type" was referred to in the title as "Myotonia Atrophica". They discuss how the condition had been described under various other names but explain their own term, which they acknowledge was "first used by Rossolimo, because it serves shortly to describe the salient features, and because it is the name under which several authors have since described this condition" (1909, p. 187). The recognition of the potential for confusion and the need to have a single name was identified and today, although the majority of literature refers to myotonic dystrophy (DM) other terms are still in use. The abbreviation comes from the Latin title Dystrophia Myotonica but the Myotonic Dystrophy support group is called the MDSG. Batten and Gibb (1909, p. 191)

thanked the doctors for their permission to describe the patients they present in their paper and referred to how they "were shown" before groups such as the Neurological Society as interesting cases. In this way the medical details of significance became the intellectual property of the researching clinician and the focal point of interest became the condition and its classification.

The history of DM, since it was recognized as a distinct category of signs and symptoms, shows how it moved from broad classification of a distinct condition based on clinical observation to an ever more detailed exploration of this classification. Initial work was carried out in the hospital or clinical setting. Later work involved the clinical researcher working within the clinic and also building up a more detailed picture through seeking out families with DM in the community. The principle of a doctor developing a specialist interest in a condition prevailed.

Methods used in the assimilation of knowledge of DM were characterised by the medical viewpoints of the time as well as being shaped by developing technologies. Around the time of Steinert's classification and into the 1920's DM studies reported in the Journal of Nervous and Mental Disease, Neurology and Brain highlighted the condition as one of neurological interest. The focus on refining classification predominated with articles on cataract, muscle and digestion. The presentation of case studies as a method of classification continued, with an emphasis on cases that would fit the category or those which challenged the classification (Maas and Zondek 1923).

The developing technologies of x-ray and microscopy influenced the ways DM was investigated. This was reflected in studies at histopathological and cellular levels. The predominant epistemology of medicine and science grounded in empiricism was apparent in studies relating measurement and statistics to aspects of the condition. Studies such as those measuring the brain indicated broader concepts of disease construction and were particular to their time, such as associations between anatomical configuration and traits such as personality and IQ (Maas 1938). Much scientific investigation was based on hypotheses that were subsequently rejected.

Analysis of the DM medical records for this thesis showed that considerable information was collected in the notes as a data gathering exercise, even in the apparent absence of related hypotheses.

> *It is also interesting that x and her mother are described as having an inordinate liking of salt.*
>
> *Physician to geneticist 1978*

Relics of past projects traced the amount of information considered and tested through a process of exclusion as well as inclusion whereby knowledge was gained incrementally by new discovery but also modified by what was disproved. This information was generally located in the clinical arena. The study of DM remained primarily driven from knowledge gained through clinical observation and then investigated by scientific methods.

Case studies further refining classification and extending DM knowledge to what would eventually become several distinct manifestations of the condition continued. The 1940s also saw the publication of studies looking specifically at DM as a genetic and familial condition. The statistical interpretation of clinical conditions prevailed with authors such as Bell (1947) framing their arguments numerically.

The increase in publications during the 1950s and into the 1960s marked a time of increasingly sophisticated focus on endocrinological manifestations of DM, continuing classification, and studies at the microscopic level such as histopathology and the gross anatomical level such as measurement of the skull and diaphragm (Caughey 1952). The application of knowledge to clinical management was reflected in studies on the dangers of anaesthesia and the refining of diagnostic muscle tests. The gradual emergence of the category of congenital DM continued. The influence of the survey, as a way of gathering information at a population level, was evident (Lynas 1957).

The wide variety of journals in which articles appeared, and their geographical locations, indicated how communication was changing medicine and the increasing technology being brought to bear on investigations. According to Harper (2005) the discipline of clinical genetics emerged towards the end of the 1950's and this was reflected in the appearance of journals specifically related to professional practice as well as research.

By the 1970s clinical assessment was combined with the technology of the slit lamp examination to detect cataract signs, the electromyograph (EMG) to detect the characteristic myotonic electrical discharge in the muscles and immunoglobulin measurement to estimate diagnostic status (Online Mendelian Inheritance in Man). Bundey et al. (1970) emphasised the importance of clinical expertise, with the technologies providing additional information.

The 1980s and 1990s were marked by the new epistemology of genetics, transformed by the discovery of the hereditary role of DNA in the 1950s. Articles refining the methodology of gene discovery showed a progression from linkage studies to gene isolation and characterisation. The clinical implications and management of DM continued to be investigated but the profile of DM was increasingly dominated by its classification as an exemplar of a particular type of inherited condition (Harper et al. 1972).

The increasing sophistication of genetic techniques that resulted in DM gene isolation was matched by increasingly sophisticated clinical knowledge, which had partly come about through the focus on clinical research. The incidence and character of cardiac involvement for example, in the 1980s and 1990s, resulted in the integration of cardiac monitoring as part of DM management (Hawley 1983; Tokgozoglu 1995).

Ideology

At the turn of the twentieth century the inheritance laws postulated by Mendel were rediscovered. As Harper (2006, p. 5) noted "... writings on the medical aspects of inheritance ... stretch back not only to the beginnings of medical genetics in 1900 but a century earlier thanks to the interest and careful reports of physicians of these conditions".

The social context in which the classification work of Steinert was being carried out in the early 1900s was one of great change and development of new ideas about the future of society. At the end of the nineteenth century the Industrial Revolution, the mobility of work forces around Europe and the United States, and the germ theory of

Pasteur and Koch had changed society radically. Medicine was organised around the clinic or hospital, with the rich being treated at home. Advances in thinking about hygiene informed the training of doctors and nurses, and the diseases being treated appeared primarily as a result of the social changes brought about by factory work and large numbers moving to poor housing conditions in cities. Vaccination and mass scale treatment resulted from the introduction of the earliest public health policies.

However whilst concern for the poor had been galvanized by the development of trade unions, leading to parliamentary action, there was also a growing sense of unease at the perceived degeneracy of the poor. In the 1800s French physicians and alienists, the early term for psychiatrists, introduced the concept that familial degeneration could occur over generations and stated their hypothesis that certain families had characteristics responsible for their decline (Lachapelle 2007). Foucault (2001, p. 269) put this into a social context through linking medical practice with moral authority, whereby the figure of authority in asylums was "the apotheosis of the medical personage".

In the UK the work of Galton and in the US of Davenport gathered credence as, fuelled by the rediscovery and reinterpretation of Mendel's Laws, science was harnessed in a program of social reform. Darwin referred to the tendency for some hereditary diseases to appear at a younger age in subsequent generations whilst Galton applied the principle to society and the way reproduction could be manipulated to encourage certain valued parts of society to have children "while those who would be a drain on society should be encouraged to have fewer children or to emigrate" (Friedman 2008, p. 20).

DM belongs in this context, as its classification and the controversy about its inheritance pattern were part of the argument. Of the two defining characteristics of DM, the characteristic muscle sign of myotonia and anticipation, myotonia gained credibility in a reasonably straightforward manner through clinical observation and examination, whilst the concept of anticipation was contested through the twentieth century. The way that certain diseases such as DM had a tendency to become more

severe in later generations had been clinically observed but the concept of an underlying biological mechanism was fiercely controversial (Tramonte 2005).

By 1905 Nettleship in Britain named this concept anticipation and began the modern debate, which had been visible during the early twentieth century through the classification work of Steinert and others, and became visible again in the work of Lionel Penrose in 1948, which set out to prove that the concept of anticipation had no basis. Geneticists such as Karl Pearson used the trend in medicine towards statistical analysis to challenge the biological existence of anticipation (Friedman 2008; Tramonte 2005).

The social significance of anticipation could be found particularly in the views of psychiatrists where they had a direct impact on practice. For those believing in anticipation for conditions such as schizophrenia and DM, it represented nature's way of bringing an end to degenerative illness within a family as it would eventually cease to be reproduced, whilst those opposed to the concept believed that it was necessary to sterilise groups seen to be at risk of passing the genetic condition to future generations.

Penrose is associated with the birth of modern genetics and the dissociation from eugenics. However he was also responsible for discrediting the concept of anticipation which he saw as a hypothesis based on incorrect methodology, rather than an explanatory mechanism with a biological basis. He used DM to explain researcher selection error and allelic modification as possible causes of the apparent increase in severity through generations of the same family, and dismissed the phenomenon of anticipation as an artefact of ascertainment (OMIM, p. 34).

The gradual disappearance of anticipation from textbooks, despite increased training and professionalisation, led to newly qualified generations of clinical geneticists opposing or being unaware of the significance of anticipation, although older geneticists trained between the world wars, and practising physicians, neurologists and psychiatrists continued to use the theory as a way of explaining cases seen in practice (Friedman 2008). Once again DM would be used as an exemplar of a particular scientific reasoning. The neurologist Howeler, in his 1989 study of DM,

raised the concept of anticipation as an explanatory mechanism based on a detailed analysis of Penrose's original work, and his own clinical studies on families. Although the evidence was compelling it took some time before it began to gain credibility, in part due to the resistance of geneticists. The influence of Penrose, benign and visionary in many ways, continued to occlude judgement in this specific area. In the UK Dyken and Harper (1972) referred to the role of anticipation, amongst other possible factors influencing the maternal link to congenital DM. Harper was primarily responsible for disseminating Howeler's work to the clinical genetic community and giving it credence. The eventual discovery of the DM gene showed the biological and molecular basis to support clinical observations made decades before.

> The recent discovery of inherited unstable DNA sequences, first in fragile-X mental retardation and now in myotonic dystrophy, not only confirms that anticipation indeed has a true biological basis but provides a specific molecular mechanism for it; this discovery can explain many of the puzzling anomalies in the inheritance of myotonic dystrophy...
>
> Brook et al. 1992

The profile of DM and the way it was part of the debate of its time, being used to "prove" both sides of the argument, gave an interesting perspective to facts about the DM which are taken for granted today and to the way dominant discourses shape discovery.

The controversy over anticipation in DM ended in 1992 with the discovery of the unstable triplet repeat mechanism whereby the mutation is an expansion in the number of CTG repeats in the DM gene on chromosome 19. An increase in the number of repeats is associated with an earlier age of onset and greater severity of manifestation.

> Anticipation in myotonic dystrophy: new light on an old problem.
> The concept of anticipation, the occurrence of a genetic disorder at progressively earlier ages in successive generations, has been debated from the early years of this century, with myotonic dystrophy as the most striking example.
>
> Harper et al. 1992

With anticipation now validated through scientific proof it reappeared as an explanation for the experiences of families and the observations of clinicians. The sociological implication for this new classification where "Significant change in the definition of the disease in that network will have far-reaching effects on the

definitions that are sustained elsewhere" (Atkinson et al. 1997, p. 123) was recognised and can be seen through networks such as the DM patient support group.

> This pattern is called anticipation. This often leads to all three forms of the disease being present in one family. The severity of the disease increases with each generation so that the family may contain a minimally affected grandparent, a classically affected parent and a congenitally affected child.
>
> MDSG website 2009

Interviews with family members for this thesis highlighted the dynamic nature of the language used to describe DM and the attribution of independent agency to the gene. This was reflected also in scientific descriptions such as behaviour being attributed to "a deranged section of chromosome" (Pryse-Phillips et al. 1982, p. 589). The transmission of DM through the mechanism of anticipation was also described in dynamic language in the publication outlining gene discovery and in the clinical situation.

> The unstable nature of this region may explain the characteristic variation in severity and age at onset of the disease.
>
> Harley et al. 1992

From genes to proteins

The DM molecular diagnostic test enabled diagnostic certainty, although the practice of predicting prognosis based on the number of repeats in the expansion was discontinued after a short time, due to the unpredictability of the phenotype at an individual level.

The experience of DM for families continues to be characterised by uncertainty, although since the advent of the gene test this has changed in emphasis from diagnosis to prognosis. The disclaimer on DNA reports at the Institute highlights the continuing need for expertise and awareness of relationships and dynamics within the family, and the dependency of the laboratory test on the clinical setting.

> *Results are dependent on samples being correctly labelled and family relationships as indicated*
>
> *DNA report in DM medical record*

The influence of earlier historical perspectives, such as the study of the "normal" against which alternatives such as ill health and deviance could be measured, persists in contemporary literature as a way of continually refining classification.

> This study indicates deviant personality in classical DM-1 regarding temperament and character, both in comparison to healthy controls and to patients with other muscle disorders with no known brain disorder.
>
> Winblad et al. 2005

There has been continuing research into the significance of repeat size to phenotypic manifestation, for example asserting correlation between repeat size and difficulty recognising key emotions in others (Takeda et al. 2009). Research into aspects of character and personality in the DM phenotype has been apparent since first classification. Fischer (1923, p. 175) wrote, "The typical symptoms are deficient mental development without gross intellectual disturbances, a certain lack of interest and stupidity, a distrustful nature, emotional coldness, lack of ordinary friendliness, distaste for work and lack of trustworthiness". DM has an extremely variable presentation, and includes people with learning difficulties in addition to those who are mildly affected. Contemporary studies focus on biological explanations such as from neurology or endocrinology to explain "the general lethargy, apathy and lack of drive in these patients" (Olsson 2002, p151). The classification of DM continues to be marked by difficulty explaining its variability, and the association between measurable aspects such as cataracts and cardiac function and less definable aspects such as lack of insight or drive. Much research on the cellular basis of DM and potential treatments is focused on the roles of the DM protein and messenger RNA and laboratory work on therapy is still in the earliest stages.

Chapter Two

Bodies of Work: Literature, Context and Background

Introduction

But sadly, it [DM] affects the heart and I'd lost ...three of my uncles had heart attacks...I mean my youngest uncle, when he was only about ten or twelve years old he had very, very bad symptoms of dystrophy – you know – his feet, hands, everything just went and in the end they had to give him a feeding tube because he couldn't swallow and eventually he died of a heart attack...so the other two died of heart attacks, but they were both affected by the dystrophy as well. My uncle had the cataracts and he couldn't get around that well...he was quite tired. They also said you'd notice baldness and they all went bald (laughter)...only one's got his hair and he's quite proud of that, all the others had gone quite bald and – you know – I noticed that they looked slightly different and they both died of heart attacks, but my mum now is very ill – she's in hospital...She went into depression...she wouldn't eat and just starved herself, and she has now...a week ago...had a coronary and they're worrying about it now because they think all this might be connected to the dystrophy.

Maria aged 46 DM family interview

Publication of the DM gene discovery in prestigious scientific journals led to media attention and to later sociological research into the nature of scientific collaboration and competition (Brook et al. 1992; Harley et al. 1992; Atkinson et al. 1997). There has been considerable research in the area of scientific discovery and scientific discourse (Latour and Woolgar 1979; Collins and Pinch 1982; Gilbert and Mulkay 1984; Batchelor et al. 1996; Atkinson et al. 1998). However there is less research into the effects of genetic discovery on the everyday work and experiences of patients and professionals (Hedgecoe 1998, 2001; Kerr 2004; Featherstone et al. 2005).

Concern about the impact of the new genetics, public understanding of new technologies, professional discourse relating to scientific and medical knowledge, reproductive implications, and perceptions of disability and impairment have been the subject of social science commentaries (Oliver 1990; Parsons 1990; Haraway 1991; Lippman 1992, 1994; Wynne 1992; Parsons and Atkinson 1993; Swain et al. 1993; Shakespeare 1995, 1998; Kerr et al. 1997; Hallowell 1997; Barnes et al. 1999; Katz Rothman 1998; Cunningham-Burley and Kerr 1999).

Wainwright et al. (2006, p. 2052) wrote of the importance of "scientific research from the bed to the bedside" to a modern biomedical society and asserted "there is currently a dearth of social science research on the interaction between the laboratory and the clinic". This echoed the work of Batchelor et al. (1996, p. 248) who asserted that too little was known about how "innovative knowledge" becomes integrated into clinical practice and called for research to take "the processes of discovery into the various domains of their production and reproduction". The 1992 DM gene discovery led almost immediately to the development of a DM diagnostic blood test available through the NHS, but the excerpt from Maria captures the uncertainty that continues to characterise DM in everyday life.

DM gene discovery belonged to the phase of isolation of genes for single gene conditions. Now, seventeen years later there has been a shift towards investigating gene function. The expectation of treatment following isolation of the gene has not yet materialised, and paradoxically a legacy of the phase of single gene identification has been to illustrate how complex the interrelationship is between genotype and phenotype. Leach Scully (2008, p. 801) recommends that geneticists "craft a different popular narrative" to convey this complexity.

The incorporation of gene discovery into a diagnostic gene test and the incorporation of the knowledge of the condition into a DM management protocol moved gene discovery from a research into a service context. The gene became part of the everyday work of the clinic, impacting directly on the diagnostic experiences of families and the professional practice of clinical and laboratory staff. In this context the experiences of those people who are directly implicated through these new technologies has been recognised as essential to the evaluation of their impact (Kleinman 1988; Cunningham-Burley and Kerr 1999).

The significance of the cultural context in which work is performed and skills developed was central to the analysis of the medical records in this thesis, reflecting Freidson's assertion (1988, p. xi) that the social value of its work is as much a function of its organization as it is of the knowledge and skill it is said to possess."

The changing context of medical practice, with focus on evidence based practice and measurable outcomes, has increased bureaucratisation and also formalised the nature of clinical medical research (Cochrane 1972; Guyatt et al. 1992; Sackett et al. 1996; Eddy 1995; 2005). This was reflected in the Institute, and exploration of the impact of the DM gene discovery took place in a setting both physically and institutionally different to the one in which the research began. Exploring the practice and experience of DM over three decades highlighted what has changed and also those issues and practices that have not, despite technological advances.

Social Science

Scientific discourse

Following the isolation of the DM gene a team of sociologists published work on the significance of the discovery (Batchelor et al. 1996; Atkinson et al. 1997; Atkinson et al. 1998). This work addressed the nature of collaboration and competition, and the career of a medical discovery in genetic research. The social processes integral to the validation of a scientific discovery, and the significance of the retrospective account in constructing the story of a discovery were examined. DM gene isolation was interpreted sociologically at the time of discovery in a way that elucidated the then revelatory aspects of scientific culture (Batchelor et al. 1997). The DM discovery was interpreted through exploration of how the scientists constructed their own accounts of discovery, rather than looking for a single account. This reflected the focus within discourse analysis "not to furnish definitive readings of texts, but to identify the recurrent structural features of participants' discourse and to describe how scientists accomplish their own reading" (Mulkay et al. 1983, p. 200).

Recognition of the importance of linguistic repertoires in scientific and medical culture shaped the development of methodology focused on discourse, as did the growing recognition of the ubiquity of narrative as a way of constructing meaning (Latour and Woolgar 1979; Bury 1982; Charmaz 1983; Williams 1984; Gilbert and Mulkay 1984; Kleinman 1988; Riessman 1990a, 1990b, 1993; Atkinson et al. 1997; Frank 1997, 1995; Ettore 1999; Hedgecoe 1999). Discourse analysis focused on describing how scientists' accounts are organized in ways which portray scientists'

actions and beliefs in a variety of specifiable and contextually appropriate ways" (Mulkay et al. 1983, p. 196).

Sociological focus on the culture around scientific discovery as a source of insight into institutional practice developed from the ethnographic work of practitioners such as Latour and Woolgar (1979). Halliday (cited in Gilbert and Mulkay 1984, p. 6) drew attention to the importance of context and the way "language comes to life only when functioning in some environment". This took account of the variable and sometimes contradictory statements apparent in versions of events. In "Pandora's Box" Gilbert and Mulkay (1984, p. 57) drew on Halliday's use of two linguistic registers to identify two principal interpretative repertoires as "a first step in making sense of the ordered variability of scientific discourse". The empiricist repertoire is used to "construct texts in which the physical world often seems literally to act and speak for itself" (Mulkay et al. 1983, p. 197) and conveys the inexorable progress of science. This contrasts with the contingent repertoire referring to contingency factors such as luck. Conflict between the two repertoires is resolved by use of the Truth Will Out Device (Coffey and Atkinson 1996). They make use of Halliday's (1978) observation that language is always used in relation to the environment in which it is functioning. This focus followed from the observations of the differences between formal scientific literature and informal scientific discourse (Latour and Woolgar 1979; Gilbert and Mulkay 1980; Collins and Pinch 1993).

Gilbert and Mulkay challenged the notion that the researcher could provide a definitive representation of the words of scientists. They argued that accounts were context dependent speech acts, and a traditional analysis of "what really happened" did not allow for a more diverse range of analytic interpretations to be represented. According to Gilbert and Mulkay (1984, p. 2) these attempts at definitive accounts "are unsatisfactory because they imply unjustifiably that the analyst can reconcile his version of events with all the multiple and divergent versions generated by the actors themselves".

Meaning in practice

The "black box" metaphor has been used to convey a perception of science as value-free and infallible. Early studies of science such as those pioneered by Popper and

Kuhn were "uncritical towards the core concepts of scientific rationality, objective truth, and logical positivism (Franklin 1995, p. 167). This was challenged by the development of the sociology of scientific knowledge, characterized by relativism and located in social constructionism (Collins 1983; Latour 1987). The development of discourse analytic and laboratory ethnographic studies emphasised the socially constructed nature of scientific practice. Practitioners of linguistic approaches and cognitive historians recognized "the interrelatedness of discourse and institutional life" (Nersessian 1997, p. 21; Fairclough 1995; Hyland 1997).

Kerr (2004, p. 3) observes how there is an attraction towards the novel and the new in popular culture and research but states that this should not be at the expense of "traditional, long-standing or apparently mundane aspects of science and medicine". She states that genetic technologies are taken for granted in sociological research and that "professional practices remain largely in the shadows" (2004, p. 6) and also questions the appropriateness of analysing and categorizing people with genetic conditions as patients. Pickering (1992, p. 2) asserts that, although the study of the processes and culture through which scientists practice is a valid site of sociological enquiry, "while science has always commanded a considerable audience, scholars have traditionally shown little direct interest in scientific practice". The scientific narratives reported in this thesis were embedded in the reconstruction of the practices, processes and relationships integral to gene discovery.

Analysis of the DM medical record archive for this study illustrated the key contribution of the families themselves and the relationships developed around a shared, negotiated and sometimes contested discourse of research and service between families and researchers (Latour and Fujimara, cited in Pickering 1992, p. 13). Analysis brought practices, processes and relationships to the fore rather than looking for definitive accounts. Collins and Yearley (cited in Pickering 1992, p. 18) indicate the importance of looking at the use of a particular position rather than the meaning. This echoed Mol's (2002, preface) observation that "Attending to enactment rather than knowledge has an important effect: what we think of as a single object may be more than one". Enactment of specific practices reflects embedded knowledge, and the significance of what actors are doing as well as saying. DM was made visible through the way families and professionals spoke about it, but also through the practices surrounding it in everyday life and work.

While narrative analysis has been used to explore the communication of science in education, analysis of scientists' own accounts as a way of exploring dynamics and process within the scientific community and its relationship with the world beyond the laboratory remain under-exploited. Studies of the use of metaphor have concentrated on how it is used to represent genetics and how genetics is represented in education, popular culture and public understanding of genetics (Roth 1993; Nelkin 1995; Milne 1998; Roth and McGinn 1998; Hedgecoe 1999; Nelkin and Lindee 2001; Warren et al. 2001; Roth and Lawless 2002; Hellsten 2005). While there has been research into the use of linguistic devices and material practices by scientists to communicate their ideas it is more difficult to find instances exploring the way that scientists utilise metaphor to give insight into the personal meaning of these processes and interactions (Gooding 1990; Ochs 1994).

Mancoff et al. (2004) warn of the lack of understanding which can occur between scientists and clinicians in translational medicine and how this could be a hindrance to delivering the promise of biomedicine. The DM gene isolation, resulting quickly in a diagnostic test, was the culmination of two decades of work grounded in clinical knowledge through close cooperation with families with DM, and researched by a team of clinicians and scientists.

The embedding of clinical genetic research in an international framework placed the analysis of the accounts of the local DM team in this thesis in a wider network of relationships. The cultural interpretation of genetics also places it in a broad context recognised by the scientists themselves, who referred to the world outside the laboratory (Traweek 1992). The influence of networks "based on shared scientific interests rather than on geographic proximity" has been described as "invisible colleges" (Lievrouw et al. 1987, p. 220; Crane 1972). The DM collaborative network shared many similar features culminating in eventual "visibility" through scientific publications and then disbanding as allegiances were formed to research new topics.

The social life of an idea

The molecular explanation for the phenomenon of anticipation was accomplished through the enrolment of scientists, clinicians and families and demonstrated how "ideas construct social relations" (Traweek 1992, p. 437).

Anticipation was a controversial hypothesis based upon clinical observation. Although academically and scientifically unpopular and unfashionable for decades its existence continued to be described by clinicians. As a scientific concept it was rehabilitated by careful disassociation from principles linking it to social controversy but it needed to be taken up by the wider medical and social community.

Prof's awareness of the dependence of categories on a "network of related concepts and propositions" (Mulkay 1979, cited in Pickering 1992, pp. 45-46) was visible in the medical records through letters supporting and communicating the hypothesis to other clinicians and families. His studies, including surveys of the incidence and severity of DM in children, elucidated the clinical classification of congenital DM and embedded the classification of DM as an inherited condition.

The personal standing of Penrose, the "founding father of clinical genetics", combined with the abhorrence of the medical and scientific community over the association with eugenics led to anticipation being discredited for many years (Friedman 2008). This occurred despite clinical evidence to the contrary and resonates with the unequal resources available to two sides of a controversy described by Collins (1983, cited in Pickering 1992).

Gene isolation resulted in the location of a biological phenomenon and in the validation of a principle. The clinical work of building up evidence to support the claim that DM became worse through maternal transmission involved family cooperation and professional recognition. Prof and the junior doctors formed a link between the heterogeneous worlds that constituted DM. These cultural relations operated in both directions highlighting the cooperation necessary to engage families, as information about the condition was exchanged for information from the families. Clinical observations, knowledge of the natural history of the condition and unusual

manifestations within families were translated and transformed, along with the blood samples, to scientific problems to be solved.

The context in which a phenomenon is judged explanatory is influenced by its ability to "meet the needs of an individual or community" which "varies with the state of science at a particular time, with local, technological, social and economic opportunities, but also with larger cultural preoccupations" (Keller 2002, p. 5). The way a phenomenon becomes understood through different social contexts, relationships and understandings was relevant to the contesting of anticipation as a phenomenon of DM and its confirmation at gene discovery (Brown 1995, p. 37).

The painstaking work, visible in the medical record archive, of establishing a natural history of the condition based in clinical assessment, observation and expertise was coordinated with the laboratory work focusing on identifying the genetic mechanism responsible for DM. In doing so the aim was to prove the hypothesis of a molecular basis for anticipation and complete the narrative of inference from clinical observation to material scientific proof. This controversy had been enacted in public, with reputations risked on either side of the debate.

Family cooperation

A significant contribution of the local team at the Institute to the international collaboration lay in the knowledge and experience of DM built up though decades of work with families. Arksey's (1994) work on expert and lay participation in the construction of medical knowledge was relevant to the research work and the continuing expertise of DM families themselves with regard to communicating and mediating information about a poorly understood condition. Arksey (1994, p. 448) references the work of Fleck and his concepts of "thought collectives" comprising both scientific experts and lay participants. Recent research concluded that the cooperative model of research enrolment has more relevance than dominant discourses of individual benefit (Dixon-Woods and Tarrant 2009) and this was apparent in DM research where taking part was expressed as willingness to help others, generally the next generation, rather than a hope for themselves.

Altruism as a motivating factor was evident in research relating to a spectrum of medical research, and could be further divided into short term motivating factors such as helping with the research project, and long term factors such as the desire to help future generations (Hellard et al. 2001; Sample et al. 2002; Mastwyk et al. 2003; Agarwal et al. 2007; Marcantonio et al. 2008). Many of the families interviewed in this thesis referred to the fact that there was no cure or treatment for DM. They also expressed willingness to help in research, reflecting Richard's (1998, p. 293) observation that "the wish to contribute to research seems to be an important motivator for many families to submit to various procedures".

The focus on particular narrative genres such as "stories of triumph, hope and promise" (Gogorosi 2005, p. 311) were interpreted by families with DM pragmatically, where the fact that there was no cure or treatment was expressed in personal terms but the promise of future developments was related to others. This theme of pragmatism has been noted in contemporary research relating to recruitment into biobanks, where motivation to participate was explored in relation to feelings of duty, a pragmatism negotiated between the researcher and participant, spontaneity, and awareness of biobanking (Toccaceli et al. 2009). The autonomy and control of the participant was key to ethical awareness on the part of the researcher, introducing issues of responsibility in research recruitment, but also highlighting a key theme of agency and control evident in the DM family interviews (Nelson and Merz 2002).

Gaining access to more information about a condition was a practical consideration common to studies investigating motivation to participate in research (Hellard et al. 2001; Treloar et al. 2007; Slomka et al. 2008). Being kept informed about the study itself was a factor, and again this was related to agency in the DM family interviews (Slomka et al. 2008). Personal experience of a medical condition and motivation to help family members were also strong motivating indicators for research participation, as was a pragmatic attitude towards time frame with the possibility of a cure for future rather than current generations of family members (Wilson et al. 2006).

Recent research suggests that a combination of factors including altruism, wish to help family members and accessing information about a specific medical condition all contribute towards participation in medical research (Henry et al. 2006). Themes arising from the DM family interviews and the DM archive were similar and reflected interest in gaining information about DM, rather than information about scientific progress (Henry et al. 2006). Being adequately informed about research and properly consulted about consent as constitutive of both good, but also successful, research practice was evident in the DM medical record and instances where this trust was questioned resulted in anger and negativity towards the geneticist. This reflected current observations on expectations, where the nature of the activity is less important than the perception and explanation of what is required and where "the reasons that people do give [for participation] are altruistic, and quite fiercely antagonistic to any suggestion of financial or other reward (Richardson 2006, p.158).

Gene Talk

Meaning in context

Since being first described by Johannsen (1911, pp. 132-134) as "nothing but a very applicable little word" the use of the word gene was a pragmatic and utilitarian choice. It is in the association of concepts with the word, and the linguistic persuasiveness of these concepts that the power of the gene can be traced. The ongoing debate over the accurate definition of what a gene actually is has been referred to as a "dialectic of structure and function" (Griffith and Stolz 2006, p. 499; Reydon 2009). The term "gene" has been challenged as outmoded and unreflective of contemporary scientific discourse (Keller 2000; Keller and Harel 2007).

The metaphor of language has been used to describe the gene itself as either a boundary object such as a code, blueprint or text, or as an agent of change capable of traversing boundaries (Lippman 1992; Hedgecoe 1999; Lewis 1999). According to Franklin (1995, p. 178) "language is increasingly the model for genes, understood also in terms of maps, codes, information systems, and switches". The influence of metaphor in popular culture has been recognised alongside the changing metaphors that reflect new information and a desire to shape or to reflect changing contexts in which information is being received (Van Dijk 1998). In this way the idea of the gene

as text has moved onto more contemporary metaphors such as that of a computer programme (Gerstein et al. 2007).

The influence of popular culture in shaping understandings of technologies such as genetics has been documented, and it is in this context that the importance of exploring the language and metaphor used by people with DM is seen (Haraway 1997). Metaphor can open up areas of shared understanding or communicate alternative perspectives (Maasen and Weingart 2000; Hellsten 2005). However this is dependent on audience and on questioning "whose point of view certain metaphors promote" (Hellsten 2005, p. 295). The concept of the gene in clinical genetics is a key part of clinical discourse and language is central to communication between families and the geneticist. The medical records document how this is accomplished through both words and diagrams in letters to families after the genetic consultation. Metaphors used in the DM family interviews reflected current thinking of the gene in terms of structure and function in that qualities were attributed to the gene regarding character and agency.

Media use of metaphors to publicise scientific issues has contributed to the public understanding of genetics but conversely the reiteration of particular linguistic devices can result in over emphasis on particular aspects of genetic conditions (Keller 1995; Kitzinger 1999; Kitzinger et al. 2003; Kitzinger et al. 2005; Petersen et al. 2005; Kitzinger et al. 2007). The appropriation of metaphors to link scientific discourse with the popular media indicates their function as boundary objects but also how they are "constitutive of certain views of the world" (Hellsten 2002, p. 3). These views may serve to further complicate the lives of people with DM through reliance on stock imagery reflective of rather than challenging existing cultural discourse.

Fox Keller (2002, p. 7) asserts that "genes display neither the stability nor the clarity expected of the explanatory elements upon which the physical sciences have come to rely". This instability and unpredictability has been apparent in research relating to DM as metaphorically associated with control and deviance, further conceptualising DM as outside of the accepted norms (McGuffin et al. 1994).

The utility of metaphor as a way of actively shaping explanation of scientific concepts, and conveying and interpreting explanation is evident within the scientific community and the health professions (Jenny and Logan 1996; Nelkin 2001; Hellsten 2002; Cooper 2007; Berdes and Eckert 2007). The adaptive requirements of accessing language to convey meanings that may change over time necessitate the use of metaphors that may be "both entertaining and research-stimulating" (Avise 2001, p. 86). The influence of the changing context of funding for science in addition to the necessity to present research in the best light has contributed to narratives of hype according to Hessenbruch (2005).

The influence of "gene talk" (Fox Keller 2000, p. 141) on acquiring funding for research on DM at the Institute could be traced through the personnel contributing to the medical records. The junior doctors contributed to both research and management of the condition and at the most intensive phase of DM research the DM clinic had a multidisciplinary team involved in service. The impact of a subsequent decrease in DM research could be seen in the diminished personnel involved with the DM clinic.

Meaning and metaphor

Critiques of the degree of pervasiveness of the medicalisation hypothesis are similar to those in the geneticization debate .The argument is encapsulated in debate over the meaning of genetics conveyed through metaphor. Hedgecoe (1998 p. 235) asserts that the use of the "blueprint" metaphor and description of "the gene as cultural icon" are located in polemic and the assumption that the public will interpret the metaphor in the same way as it is postulated rather than "convincing empirical evidence". A need to focus on small studies examining the impact of geneticization on single disorders as a way of testing claims echoes Kerr's (2004) observation that evaluation must recognise the experiences of individuals who are directly involved with medical genetics. Hedgecoe's (2001, p. 307) definition of geneticization as "making an explicit link between a condition and a stretch of DNA" allows all aspects of genetics as it is experienced and practised to emerge.

Cunningham-Burley and Kerr (1999, p. 662) assert, "Medical sociologists too must carefully negotiate the twin poles of biological and social determinism". Extreme interpretations of the medicalisation and geneticization hypotheses risk marginalising

people with DM whose experiences of diagnosis, medical management and reproductive risk offer insight into alternative representations of living with an inherited condition. The portrayal of society in general, or families with genetic conditions in particular as passive and unable to process information or articulate viewpoints has been challenged in relation to both medicalisation and geneticization (Williams and Calnan 1996; Hedgecoe 2001, 1998).

The involvement of clinicians in areas beyond the immediately medical has been classified as part of medicalisation in that it extends the power and legitimacy of the clinician beyond the medical and into the social domain (Conrad 1992, p. 216). However where "medical explanations were mobilised to enhance the coherence of the patient's experience of symptoms, patients found medicalisation to be helpful" (Broom and Woodward 1996, p. 358). Cunningham-Burley and Kerr (1999, p. 660) note the "social values" apparent in the practice of some geneticists engaged with the implications of the new genetics. The DM medical records documented communication over decades between geneticists and agencies such as social services, employers, and schools, in addition to other health professionals. Correspondence related primarily to accessing services or resources and to providing official proof and explanation of the DM diagnosis. The need to provide legitimisation to employers or resource providers shows how the classification of DM itself was subtly changed as it crossed boundaries of medical and social worlds, and how the work of the DM clinic remained connected to the implications of DM for everyday life before and after gene discovery.

Focusing upon "'patients' experiences beyond the clinic gives but a partial perspective on the ways in which people live with genetic disorders" (Kerr 2004, p. 101; Richards 1993; Lippman 1994). According to Parsons (cited in Clarke and Parsons 1997, p. 257) medical genetics has been evaluated within a context of whether it is acceptable or not and a more contemporary approach necessitates engaging with the epistemology of the social scientist to ascertain their framework of reference and then develop a "personal understanding of the relationship between genetics and culture." Another aspect of this involves engaging with the practices and events that constitute medical genetics as families and professionals experience it. The need for medical genetics, especially with rapid advances in technology, to be

evaluated through psychosocial research has been expressed by geneticists within the field (Harper and Clarke 1990; Harper and Clarke 1997; Gill and Richards 1998).

In their review of medical and scientific discourse related to the new genetics Cunningham-Burley and Kerr (1990, p. 653) refer to terms such as "dreadful disease" and "plague" used by James Watson and contrasted this with the more "circumspect" language of some clinical geneticists. The DM medical record archive contained some early letters from family members where they wrote directly to Prof asking for help and offering assistance to research "this dreadful disease." The variety and complexity of experiences, situation, decisions and perceptions within family records and interviews illustrates the difficulties of trying to attain a single account or explanation of the impact of gene discovery, but indicates some broad themes. These necessitated resisting the definition of a person by their medical condition while also accepting that it is in the context of having DM that this research was being carried out.

Patient People

Defining issues

As DM is an incurable and degenerative condition consideration of concepts of health and illness, chronic illness and genetic disease informed decisions about the context of this thesis. In their meta-study of the experience of chronic illness Thorne et al. (2002, p. 443) asserted, "the very notion of chronic illness is itself contested" and that populations studied rarely included those with articulation or verbal difficulties and concentrated on the ambulatory

According to Charmaz (1987) people with chronic illness wish to be seen as people first, rather than as patients. The concept of identity as challenged by chronic or degenerative illness was explored and the narration of how personal biography was altered by diagnosis echoed previous sociological work (Charmaz 1987). Bury's (1982, p. 167) conceptualisation of "Chronic illness as biographical disruption" focused on changes in perception of personal biography due to illness, and the impact of relationships and resources on the way illness is experienced. The "loss of self" (Charmaz 1983, p. 168) caused by chronic illness and the subsequent impact of this altered perspective in restricting possibilities highlighted how illness is

experienced in everyday life rather than a medicalised perspective. The strategies used to narratively reconstruct personal biography in response to chronic illness illustrated the way the meaning of illness was incorporated into understanding and experience, rather than solely focusing on problems caused by the illness (Williams 1984).

Kleinman (1988) notes how the doctor and the clinic construct the case and in so doing transform illness into disease. The association between agency and the role of a patient "which is so redolent with the sights and smells of the clinic and which leaves an afterimage of a compliant, passive object of care" (Kleinman 1988, p. xiv) was relevant to the family interviews in the way that there was resistance to being defined as a patient. The struggle to maintain agency was a key theme of the family interviews. This extended beyond the clinic and related to coping with the physical deterioration associated with DM, and accessing the resources, both material and emotional, necessary to maintain roles of everyday life.

The conflict between being continually placed in the role of patient to gain resources, and the wish not to be defined by a diagnosis has been addressed by Scambler (1997, p. 173) in his analysis of the stigmatizing potential of diagnostic labels, which can "come to dominate the perceptions that others have of them and how they treat them". The possibility of challenging stigma and stereotyping has been recognised, as has the way it may be easier for a person with a disability to conform to the expectations of others (Higgins 1980, cited in Scambler 1997, p. 175). A feature of DM is excessive tiredness and muscle decline necessitating increased effort to accomplish everyday tasks. The impact of physical tiredness on attempts to alter expectations of documented characteristics such as apathy should be recognised as part of how the material body directly impacts on agency.

The difficulties faced by people with disabilities in escaping "the coercive power of attributed identities" where there is a failure by members of society "to recognise any aspect of their public identity other than their disability" (Taylor and Field 2003, pp. 126-127) was narrated, but so too was the difficulty in validating a little known condition. Family members focused on the manifestations of DM in relatives further complicating the delineation between condition, role and individuality.

In her "Shifting Perspectives Model of Chronic Illness" Paterson (2001, p. 21) describes people with chronic illness moving between states where the "illness" is either foregrounded or in the background. The necessity of having to present an identity as a patient was observed by carers who constructed accounts of wanting to stay positive and optimistic about the future but found that a more negative approach was necessary in order to attain the necessary resources to maintain independence for longer. Resources were central to the DM narratives, reflecting other studies indicating how aspects of chronic illness and disability are social rather than medical in their consequences (Crossley 1998; Larson 1998).

Functional definitions of health as being able to cope with daily activities and negative definitions such as not being ill were commonly invoked (Pill and Scott 1982; Blaxter 1990). This reflected the work of Stuifbergen et al. (1990) who found that descriptions of good or excellent health did not corroborate with actual physical function, and this was attributed to an adjustment in expectations rather than a false view of reality. The perception of the person themselves and the context of the condition, particularly family attitude to DM, as important influences on behaviour and attitude reflected the many possible representations of DM beyond the clinic (Zola 1966).

Concepts of disability and impairment were relevant to the difficulties expressed by families in maintaining roles of everyday life. The impact of DM on the body and resulting deterioration in function was recognised but so too was the impact of the social perspective in terms of accessing necessary assistance to maintain an adjusted version of everyday roles. The debate between medical sociology and disability studies was explored with essential viewpoints from both perspectives being recognised (Oliver 1990,1991,1996; Shakespeare and Watson 1997; Williams 1999; Bury 1997, 2000; Albrecht et al. 2001; Finkelstein 1997, cited in Rapley 2004, p. 63; Shakespeare 1995, 1998b, 1999, 2006; Thomas 2004, 2007). Mediation of the work of medical sociology and of disability highlighted the possibility of integrating the dual nature of social process and bodily degeneration in potentially limiting opportunities for those with a DM diagnosis (Williams 2006). A social relational understanding of disability, with its attendant possibility of being made manifest in a

similar way to sexism and racism (Thomas 2003), offered a practical way of recognising the physical and the social implications of a degenerative condition such as DM. However, although the content reflected themes within the family interviews, the location within a conceptual framework of disability remained problematic.

The epistemological position of this thesis viewed ability and disability as a continuum along which everyone is more or less impaired and which "can be used as a springboard for dismantling socially constructed divisions between 'the disabled' and the 'normal' (Shakespeare and Watson 2001, cited in Thomas 2004a, p. 574). This facilitated recognition of DM diagnosis and its impact on families, while recognising that the experience of DM was narrated from the perspective of everyday life.

Body, Story, Self

Narrative is a ubiquitous, powerful means of expressing personal meaning, whilst recognising the autonomy of the narrator and the socially constructed nature of the event (Polkinghorne 1988; Bruner 1990; Coffey and Atkinson 1996; Elliott 2005; Riessman 2008). The act of narration and participation in interviews was perceived as a symbolic act where the body was an expression of agency. The richness and power of the language used in the DM family interviews was notable, and reflected how "narrative makes actions intelligible to the self and others, by showing the part they play within an intentional project" (Skultans 2000, p. 9.)

Kleinman (1988) discusses the value and need for clinicians to attend to patient narrative as a way of interpreting the illness experience from the patient's point of view. This recognises the potential power of the biomedical model to treat cases rather than patients. Interest in narrative has been demonstrated in research by medical practitioners the culture and practice of medicine, and the experiences of patients (Greenhalgh 1998; Mattingly and Garro 2000; Gwyn 2002; Charon 2006). The use of narrative to give insight into organisational culture is relevant to both the culture of scientific discovery and the clinical context in which it is researched and carried out in practice (Boje 2001; Czarniawska 2006). Although families with DM resisted the definition of patient or disease many of the experiences they described have been recognised in literature on chronic illness and disability. Narrative analysis

frameworks such as illness narratives, the disruption of personal biography and the work on establishing personal meaning on the experience of illness were relevant for the analysis of the DM interviews (Bury 1982, 2001; Charmaz 1987, 1991; Frank 1995, 1997). Hyden (1997, p. 49) draws attention to the power of narratives to "give voice to suffering in a way that lies outside the domain of the biomedical model". However the reference point for the families with DM was not identification with illness or the role of the patient, but in incorporating a genetic condition into everyday life.

The body was viewed as the outward manifestation of inner intent and purpose, following the tradition of authors such as Merleau-Ponty and Sacks, and was seen as "the embodiment of who we are" (Corbin 2003, p. 258). Awareness of the body resulting from disease, injury and genetic risk places the body not just in the context of physical processes but as embodiment of the self, in that aspects of identity performed by the body may no longer be possible (Leder 1990; Hallowell and Lawton 2002). The way that social dys-appearance may lead to physical illness was recognised by Leder but not the possibility of the converse being equally applicable. According to Shilling (2005, p. 186) there is a danger of marginalizing "those people for whom the body is regularly fore grounded as an essential part of their identity" such as people with DM. The body viewed as a project or a process acknowledges the presence of the body but places it in the context of those for whom the project is a possibility rather than a necessity and again raises the possibility of documenting the dramatic and unusual rather than the challenges of those whose agency is constrained by the physical limitations of their body. The epistemological position of this thesis does not equate the body with the self but recognises that "when bodily demands conflict with desired self-presentation the individual becomes acutely aware of the divergence between body and self" (Kelly and Field 1996, p. 245). Mol (2002, p. 27) observed, "...there are ways of ethnographically talking bodies. There are good reasons to try, if only this one: that the *humane* does not reside exclusively in psychosocial matters". The DM family interviews for this study, in recognising the challenge of articulating experience due to physical limitation, and their analysis, which acknowledged the influence of the body in terms of meaning, attempted to restore agency to the way people "talked" their own bodies and experiences.

The body as the site of multiple interpretations was narrated in the DM family interviews where instances of scrutinising one's own body and those of family members was part of a process of establishing some certainty about past manifestations and future representations of DM. In this way the body itself became a temporal narrative, inscribed with both the text of the past and the future.

The body as the medium through which the intention or desire to enact a role is communicated was expanded in the DM family interviews to consider the narrating of previous roles as meaningful and representative of identity. Disrupted biographies illustrated how the present and future were narrated through the evocation of past selves (Bury 1982; Charmaz 1983; Devins 1994; Corbin 2003). Different experiences related by families were interpreted as suggesting how disability just as racism may be "grounded in culture as well as in nature" (Mol 2002, p. 18). The narrating of varying difficulties in accessing resources and information, and the differing responses within families to diagnosis were considerations that influenced coping. The construction of an illness identity has been documented, as has the differentiation made between illness and health by people who have had a medical diagnosis (Frank 2000, 1998; Corbin 2003).

The importance of the body to the work of relationships and communication has been recognised, as has the way chronic illness may result in altered relationships with the body as an expression of personhood (Turner 1996). Kelly and Field (1996, p. 247) describe how "coping with the physical body has to precede coping with relationships".

The contextual and contingent nature of interviews reflected narrative autonomy and the way "interviewees always make choices about what to divulge" (Riessman 1993, p. 52). Analysis did not seek a "true" picture of how people with DM experienced their lives but focused on exploring how people constitute their selves in everyday interaction (Bury, cited in Kleinman 1988, p. 282). Atkinson and Delamont (2007, p. 197) have expressed caution about appropriate use of narrative and the importance of an analytic focus and they warn against the dangers of assuming "privileged access to personal experience". The moral aspect to narratives and the agency they grant to presenting a chosen perspective enabled conflicting events to be incorporated into an

account (Pinder 1995). Gabe et al. (2004, pp. 71-72) draw attention to Riessman's caution about presenting narratives romantically, without interpretation, or with a redemptive overtone.

The way "personal narratives are means by which the links between body, self and society are articulated" (Kleinman 1988, p. 281) was interpreted in this study as giving recognition to all three aspects and valuing narrative as a way of reflecting them. Van Wolputte (2004, p. 254) outlined three main areas of focus as the individual, phenomological body, the social body and the body politic, and stated that what "interarticulates nature, society, and individual, are emotions". A key aspect of the narratives given by the scientific team and the family members was the emotive language used, reiterating the possibility that narrative provides a way of acknowledging the body as one of the factors that influence and impact on how experience is represented. Narrative provided a means of articulating those aspects of scientific work underrepresented in scientific discourse, and of asserting the importance of everyday life on a condition primarily represented in medical discourse. Narrative also challenges the interpretation of contemporary assertions such as that the body "is a canvas" that is "fully customizable and adaptable" (Van Wolputte 2004, p. 8) by at least providing a possibility of additional discourse from people whose experience may be different. This does necessitate practical engagement with methods such as how to facilitate interviews in physically challenging situations but the experience of the DM interviews in this thesis indicated that this was not an insurmountable issue. Booth and Booth (1997) note the inclusive possibilities of narrative for incorporating experience from people, regardless of ability, whilst offering solutions for potential practical difficulties.

The possibility that narratives may reconstruct illness as a moral occasion (Franks 1995) or that "chronic illness often crystallizes vital lessons about life" (Charmaz 1991, p. vii) has been recognised. However the reluctance for people with DM to classify it as an illness presupposed alternative interpretations of living with an incurable and degenerative condition.

The DM family narratives were not rooted in purely biological factors or pre-determined structures such as illness narratives and echoed the work of Hallowell

and Lawton (cited in Kerr 2004, p. 95) on the way biological conditions "are presented as one part of a wider complex of familial relationships and self-identities".

The way "The wheelchair, the white cane and the crutch signal bodies that do not work in 'normal' ways" (Thomas 2002, p. 64) was narrated in the DM family interviews, where the wheelchair symbolised a transition from ability to disability. Thomas (2002, p. 64) has made the comparison between meanings associated with bodily difference and with other issues of representation such as racism or sexism in her exploration of the 'disabled' body. This was carried out through engagement with the material reality of the body and contemporary debate over the place of the body in sociology (Thomas 2002). In common with other aspects of sociological thinking such as social constructionism the body and disability has been seen as the site of contested discourse. This was a reaction to a naturalistic or sociobiological approach that was interpreted as being deterministic (Shilling 2005). The contribution of social constructionism to "bring society into the body" has been most notably made by Foucault, Goffman and Turner who raised awareness of power relations and the body as social symbol. Foucault's (1973) assertion that the body is produced by discourse was evident in the way that family members became part of the discourse of DM. Kirmayer (1992, p. 341) recognised the need to integrate the "socially constructed and the bodily" and asserted that metaphor theory could make a useful contribution.

Criticism of radical forms of social constructionism focused on how it did not address the lack of agency engendered not just by social restrictions but also by the materiality of the body (Bury 1986, 1988; Shilling 2005). By ignoring the body the possibility for hearing the discourses of those with challenged bodies cannot be met. Frank's (cited in Shilling 2005, p. 86) definition of the way in which the body engages with "action tasks" addresses the corporeality of the body but references the body to social systems and discourse rather than as essential to human agency. The divergent approaches, from the naturalistic theories of Laqueur, the social constructionist perspectives of Goffman and Foucault, the body as expression of habitus in Bourdieu and the civilizing view of Elias, share a common theme of the body as the site of theories rather than as a site where access to self-expression is limited by the body itself (Shilling 2005). The body as an instrument of gender

differences, political and economic repression or capital is recognised, as is the possibility for spoiled self-identity through inadequate performance. The body as an entity that cannot express the self, through challenges of physical articulation is less represented. Discourse itself may be challenged, and the body may be allocated a discourse, such as that of disability, by others rather than chosen by the self.

Narrative provides a means of reconciling the gap between the social and biological representations of the body. The DM family and scientific interviews were distinguished by the fact that, regardless of seniority or status, experiences were conveyed through the structure of narrative. The enactment of the self, through the scientists telling stories of how they built models in the laboratory conveyed role and performance, whilst the narratives of the family members directly challenged the physical restrictions of the body affected by DM through the strength of the language used.

The iteration between the body, emotion, and response from others was recognised by Freund (1990) and was relevant to the DM family interviews where awareness of the lack of social cues available through decreased facial muscle tone resulted in decreased responsiveness from others in social situations. Freund (cited in Shilling 2005, p. 101) describes how "emotional modes of being connect our embodied selves to social relationships in ways which fundamentally shape our ability to achieve emotional well-being". The structure and linguistic choices of the narratives offered an alternative mode of expression. However the question of audience remains, as the process of narration is democratic as evidenced by the ability of every participant in the study to produce narratives, while being heard, in both a physical and a meaningful sense, is not.

The DM Archive

Context

The DM medical records comprise a collection of documents compiled over three decades, as part of the everyday work of the Institute. The archive referred to the static place where the records were stored but the nature of these documents was revealed by considering them as texts in action. Missing records were generally "in clinic" or with the administrative or medical staff "doing letters". They circulated

from the archive to clinics and home visits, fulfilling a central role in recording DM as it intersected with clinical genetics.

The DM medical records in their everyday context were more clearly understood by referring to them as "the notes". This utilised Spradley's (1979, 1980) concept of a domain or folk symbol representing a recognisable category as it is used and accessed in its everyday context. As such the notes were part of the culture of medical genetics. Spradley's work is associated with ethnography and an immersion into a culture in order to appreciate the participants' own understanding of it. Although this thesis is not an ethnography, reference to his definition of a domain provided a framework through which to explore the significance of the records as socially situated products (Spradley1979; Mac Donald 2001). Spradley (1980) highlights the importance of contradictions or conflicts within a culture. The notes contained references also to family members who resisted the definition of inherited disease, in one instance referring to how a father had a religious cure, and in another an older generation who declined the diagnosis despite younger generations of the family coming to clinic regularly. The notes made visible the negotiation and preservation of boundaries. The capacity of a culture to exert social control was recognised by Spradley, as was the potential of documents to shape future action (Prior 2003; Atkinson and Coffey 2004).

Atkinson and Coffey (2004) state the necessity of seeing a document in context and remind that a document is not a representation of an institution or institutional practice but performs part of the overall representation. They highlight the danger of using documentary sources "as surrogates for other kinds of data" (2004, p. 58). The medical records are seen as text in a "network of action" (Prior 2004a, p. 77).

The medical records were the most representative artefact of DM within clinical genetics. Medical geneticists do not generally carry stethoscopes, lead ward rounds or wear white coats. Their identity is linked to decoding the body and its pathology in terms of its underlying genetic basis and interpretation of familial hereditary implications, through representation in words and symbols contained in the medical record. The notes had historical and contemporary relevance. Analysing them as the notes accessed the everyday way they were used by the people whose inscriptions

they contained and the shared frame of reference that contributing to the notes implied. It focused on the texts in action, as part of a network of relationships and through a cross section of records over time, allowing the factors that had influenced their development to emerge. It was evident that the notes were part of every aspect of work from administrative to clinical and looking at the notes as they functioned within their particular culture gave a context to the narrative of DM within the Institute over time. Prior writes, "without documents there are no traces" (2003, p. 165). The notes contained traces of every aspect of DM from the setting up of the department by one clinical geneticist to their function in the contemporary context of a large, internationally recognised institution. The medical records are documents "constructed in accordance with rules, they express a structure, they are nestled within a specific discourse, and their presence in the world depends on collective, organized action" (Prior 2003, p. 12).

Function

Berg (1996, p. 510) describes how the medical record performs the function of "mapping the patient...so that it matches its map" and states that the medical record is "where the inscriptions end up, are matched and rearranged, and where new inscription-yielding activities begin". The objective of analysing documents is "to analyse how they work to achieve particular effects – to identify the elements used and the functions these play" (Silverman 1993, p. 122).

However the medical records for DM differ in that the map was being defined as the records were being produced. When the records were first set up there was no gene for the condition, which was only partly understood. At present although the gene has been isolated and there is a fuller clinical picture with sub-classifications such as congenital DM, research on DM continues.

Berg (1997, p. 99) observes, "formal tools contain a predisposition to build simple robust worlds, without too many interdependencies or weak spots where contingencies can leak back in". Definitions of the function of the medical record refer to systematic documentation and case history as a chronological and neatly ordered account of a patient and their interaction with the medical system. Referring to a medical record or a set of case notes imposed order on what is in reality a messy

collection of contributions from a wide variety of actors. There were common aspects to be found in each record but the records collectively marked a period of change, discovery, interpretation and reinterpretation. They also documented changes in their own location, as successful advances in clinical genetics resulted in the speciality changing from a small department to a regional Institute with worldwide links.

Atkinson and Coffey (2004, p. 63) draw attention to the layout and the character of a document noting how "it betrays its character through various stylistic conventions". Changes over time were apparent in how the notes were put together and by whom. Later records had bureaucratic as well as medical identity, with administrative forms documenting the source of the referral for legal and financial as well as clinical purposes. Contemporary notes had dividers with sections for Family tree, Clinic sheets, Assessment forms, Correspondence, Consent and Results. This marked the way the DM clinic was practised and the notes traced this structure from its origins in the clinical examination and research of the first records. The presence of two hospital numbers on the front of each record, one for the NHS and one for the Institute, signifies the boundaries around individual medical information. Rees (1981) observes how the initial encounter with the patient on a ward is structured into a formalised account that is part of medical training. Medical genetics is an outpatient speciality but it too has its structuring activity in the form of the family tree or pedigree. By implication this involves more than the person who has been referred, known as the proband. Awareness of the sensitivity of information about extended family led to the clinical genetics records being kept separate from NHS records from the beginning of the archive. The genetic number on the front of the record serves to delineate genetic health information from any other clinical information in the NHS.

Looking at a document as an organizational entity gives clues as to "how the system – as a technology - is nested within a web of activities" (Prior 2004, p. 78). Activities as diverse as drawing the family pedigree, consulting with the neurologist, or making letters of representation from the family to social services form part of the institutional work of defining DM as a hereditary condition. Traces of these activities are located in the medical record, which also functions as a way of ordering people

and events. Where a document has evolved over time traces are left of the way these changes have occurred. Silverman (1993, p. 132) notes, "...a concern to assemble credible files may be a common feature of organizational activities". Technology is reflected in the production of the DM record as it gathered a bureaucratic identity evident in standardised forms, both laboratory based and reflecting institutional practice as part of a large university hospital (Bowker and Star 2000; Boje 2001).

The DM medical records exist in a state of perpetual possibility, in that they reframe the past, record the present and reference the future in terms of potential relationships, risk and reproduction. According to Raffel (1979, p. 43) "The record thus makes the present permanent and eternalises the event." In an inherited condition such as DM the temporal aspect of past, present and future are more than markers referencing the trajectory of management of the condition as they also serve as resources for the future.

History

The medical record as a document of analytic interest is a phenomenon of modern medicine. Berg and Harterink (2004, p. 15) traced the development of the record from being clinician centred, in that it was often a large book in which the clinician noted details of private patients, to the development of the patient centred record "fundamentally intertwined with the new shape that both the patient's body and the medical institutions acquire". They saw this change occurring in the early part of the twentieth century as new investigative procedures explored the way the physiology of the body functioned. This was a development from Foucault's (cited in Berg and Harterink 2004, p. 13) modern anatomical body where the patient was subjected to the "clinical gaze", to a focus on bodily process and function that entailed multiple opinions and investigations. The repository of these explorations was the medical record. As medical knowledge grew and different specialities concentrated on their areas of expertise the medical record reflected specific practices.

With the emergence of medical genetics there was a shift in thinking towards the underlying way the body functioned. Looking at disease in the context of heredity entailed expanding the remit of enquiry from a case history to a family history. The medical record is common to all aspects of medical care but this thesis illustrated

how the medical record of a particular condition does the work of giving identity and cohesion to the many disparate activities that make institutional practice recognizable and visible. The epistemological standpoint of the researcher was to view the DM medical record as an artefact while recognizing that access to it is privileged.

Meaning

The medical record is taken for granted in routine medical care. It is part of the apparatus of the medical consultation, often attracting attention only when it can't be found. Referring to research into the influence of the medical record on institutional practice and patient care Rees (1981, p. 55) commented, "...one could be forgiven for thinking that medicine is a purely oral discipline". However the work of the medical record as a document that constructs and is constructed by the consultation offers insight into the many activities that are occurring. The medical record is a repository of documentary evidence offering insight into the ways that "principals attach meanings to their activities and 'problems'" (Silverman 1993, cited in Coffey and Atkinson 1997, p. 5).

The focus of this thesis was to look at the underlying processes that underpin the notes and "how they work to achieve particular effects" (Silverman 1993, p. 122) rather than give either a definitive account of gene discovery and its impact, or a definition of what a document such as a medical record does. The central importance of documents to institutional life and practice has been acknowledged, together with the need to treat documents as topic and not just resource (Scott 1980; Hammersley and Atkinson 1995).

The content of the record in medical genetics is important as it introduces the particular vocabulary and characters distinguishing it from other medical specialities. The primary focus is less on the material nature of the medical record and more on the dynamic nature of the document as it functions "in action" (Prior 2003, p. 3). While the authenticity of the document is easily verified it is worth noting that documents reflect a particular agenda, and not everyone represented in the notes has equal access to them (Scott 1990). Documents are "produced in social settings" (Prior 2003, p. 26; Prior 2004) and are not only receptacles of content but also have agency.

Documents are busy and "peopled" places. They have characters or subjects and "by analysing the construction of subjects, we get to the heart of the work of the text" (Silverman 2004, p. 138). A primary question when looking at the medical records archive is who is the document for? Sociological analysis of accounts includes a "focus on the motivations and reasons behind the presentation of accounts and the influence of others (audience) in the account-making process" (Orbuch 1997, p. 456).

The DM Clinic

Classification and Uncertainty

The management of uncertainty in the everyday life of family members with DM and the everyday work of professionals' necessitated negotiation of boundaries. Boundary objects as "those objects that inhabit several communities of practice *and* satisfy the informational requirements of both of them" (Bowker and Star 2000, p. 16) served as a definition for the contextual nature of DM classification and the work of families and professionals in negotiating everyday work and practice. Boundary work between DM research and service through the DM clinic was constant and dependent on flexibility in order to achieve the current classification of DM.

The clinic was the site where reconfiguration of person and family into a DM framework took place. This was extended to the home visit and the medical records as sites where different discourses between family and geneticist were interpreted. The medical record also captured the traces of DM as interpreted by the laboratory. Classification over time moved from early representations of DM as a loose collection of signs and symptoms given greater clarity by neurological and muscle investigations, to later representation by linkage of blood markers, and to the contemporary representation as a dysfunctional gene in a specific location with specific characteristics. Prior (2004a, p. 78) writes, "...associated with each and every classificatory system is a set of practices" and notes that record keeping is of necessity selective in its observations. The DM records present an evolving picture of a condition that is directed by the geneticist.

> *...it was noticed that she had a myopathic facies with a tented mouth and was floppy. Her mother also has myopathic facies and typical myotonia*
>
> *Paediatric neurologist to Prof 1978*

The clinical picture of DM was built up and given credence by systematic focus on key defining signs such as myotonia and early cataract development. Later investigations of the heart and muscle added to this information. The family member may have been completely unaware of these and the reconfiguration of the person into the disease continued with the investigations at molecular level culminating in the definitive gene test, applicable irrespective of symptoms.

> *...x shows a typical maternal inheritance of the congenital type of myotonic dystrophy and as is so often the case the mother is relatively very mildly affected. I suspect that there are a considerable number of people in this family who may be at risk but who are not aware of it at present*
>
> *Geneticist to GP 1973*

Fleck (1979, p. 30) postulated, "Discovery is thus inextricably interwoven with what is known in error. To recognize a certain relation, many other relations must be misunderstood, denied or overlooked". In order to define the condition and further hone it down to a neuromuscular degeneration particular signs and symptoms were noted and investigated. The medical records contain information on many clinical observations and aspects of a medical history but the search for the DM gene concentrated only on some of these. Categorisation or classification as a way of ordering relations between phenomena gives insight into the structure that underpinned these activities. The "focus on the arrangement of words and sentences and things, instead of meaning" (Prior 2003, p.25) was exemplified in the work of Levi-Strauss and Foucault, while Sack's work on membership categorisation devices (MCD's) also links the purposive connections between words. Thus the category of medical genetics relates to activities such as drawing up the family tree, and the category of DM related to symptoms such as myotonia and cataracts. These categories were constantly reviewed and expanded in the notes in order to further explore the DM classification.

Before the DM gene discovery "the politics of definitions" (Conrad and Schneider 1992, p. 22) surrounding diagnosis involved ongoing research into refining the DM classification, in addition to applying clinical knowledge to making the DM diagnosis in individual family members. Classification in practice was an essential and reproducible tool for reinforcing the genetic paradigm, and professional and institutional identity. The medical records give insight into how particular phenomena were selected for investigation (Yoxen 1982; Hedgecoe 2003). The

material reality of DM was recognised but the construction of an inherited neuromuscular condition required iteration through practices recognised beyond the DM clinic. The role of professional presentations, case histories and publications in extending classification were part of the history of DM recorded in the archive.

The uncertainty and plasticity inherent to classifications that are the focus on continued research and therefore always potentially subject to change reflected DM work (Kerr 2004). Blaxter (1978, p. 11) described the "museum" of previous forms of knowledge related to diagnosis. The medical records were significant for the way in which all traces of activity remained. They captured the research projects of the junior doctors and the way incremental knowledge was applied to the clinic in addition to the DM classification. Contemporary laboratory reports and clinic letters reflected the possibility of changing knowledge and reinforced DM as a classification in progress.

Mischler (1984, p. 106) discussed how diagnosis was emblematic of the differences between the everyday and the medical world, contrasting "the voice of medicine" and the "voice of the lifeworld". The development of diagnosis and classification were iterative up to DM gene discovery. The "voice of the lifeworld" informed clinical diagnosis but knowledge of the natural history of the condition was also vital in identifying the underlying molecular basis. The embedding of service and research in the developing DM classification was preserved in diagnostic and management protocols after gene discovery. The "voice of the lifeworld" continues to be documented in the DM protocol.

The observation that "classification systems are compromises, and compromise entails debate, disagreement and dispute" (Hedgecoe 2000, p. 58) was apparent in the history of DM itself and in the different representations seen in the medical records. The necessity to present aspects relevant to differing contexts was visible in letters to social services or employers.

Classification as a balancing act involving "negotiated compromises between different groups" (Hedgecoe 2003, p. 63) was part of the wider discourse about the phenomenon of anticipation, and also the clinical management of DM as a genetic

condition. Literature relating to medical management of DM, since its classification by Steinert, involved many different medical specialities due to the multi-systemic implications. Steinert was himself a neurologist and the neuromuscular classification is still prominent. However the practice of genetic counselling distinguishes it as primarily a condition classified by inheritance

The way the clinical encounter and its subsequent, or sometimes simultaneous, documentation is ongoing, constructing DM through a variety of different sources such as clinical examination, imaging and molecular testing, reinforces the temporal ordering of DM as a trajectory. The development of protocols reflected wider changes within medical practice related to technological advance, application of scientific principles of reasoning and economic pressure of audit (Berg 1997). The DM clinic protocol tells the narrative of DM as it is medically constructed but allows for recording of the family perspective. This is in the form of a blank section of the protocol, which, in contrast to other sections plotting individual change, narrated those issues where DM as experienced by the families over time have remained unchanged.

The intersection of the medical with the social trajectory of DM could be seen here in the translation of changes in muscle function to difficulties climbing stairs, or in the excessive daytime sleepiness on work and relationships. While the issues remained similar over time there were technological applications such as drug treatment for sleepiness, in addition to the technological advance of the gene test. The potential of genetic technologies for "surveillance creep" and discrimination was evident in the medical records through communication between families, geneticists and insurance companies.

The call for small studies focused on "the seemingly mundane" aspects of care such as the medical record (Timmermans and Berg 2003, p. 108), echoes the call for studies focused on the experiences of those who are involved as patients and professionals. The construction and multiple representations of DM, from molecular to everyday life, evident in the DM medical records illustrated how different discourses as well as technology shaped the clinical encounter (Mol 2002).

Uncertainty as part of the clinical encounter has been documented, as has the way that clinicians are trained to deal with certainty (Atkinson 1984; Griffiths et al. 2005). The work of protocol, technology and discourse in managing uncertainty about DM prognosis within the clinical encounter was visible in the medical record (Casper and Berg 1995). The central role of language and the uncertainty associated with communicating concepts related to the gene has been noted. A letter to the family after consultation was part of practice from the beginning of the DM archive and captures the way uncertainty is shaped into accounts that integrate the possibility of future knowledge changing current information. In this way uncertainty becomes part of the definition of the condition and part of everyday practice. The value of written summaries has been evaluated (Hallowell and Murton 1998) but the DM archive provides insight into the way taken-for-granted activities evolved. Through iteration in the medical record generations of training geneticists wrote clinic letters to families that became institutional practice.

Technology

Technology within reproductive decision-making has been addressed by disability, biosociality and feminist discourse in addition to genetics (Parsons and Atkinson 1993; Denny 1994; Katz Rothman 1998; Rapp 2000). The study of technology in action allows the gene test to be seen through practice (Timmermans and Berg 2003). The implications of an individual test on the wider family can be seen as the test results may include or exclude extended family from a potential DM diagnosis. The application of this technology in the clinic results in diagnosis being explained in individual terms and as part of a wider discourse of the whole family. In this way the gene test becomes part of a process beyond the boundaries of the technology itself.

Uncertainty is still present, in relation to prognosis but also occasionally in unusual presentations of the condition (Latimer et al. 2006). The clinic as the site where the boundaries of laboratory, family, and clinical judgement interact was not marginalized by the advent of new technology in the form of the gene test. It has continued to function "as a site for knowledge production" (Latimer et al. 2006, p. 624) in relation to evolving clinical management. The new technology was incorporated into a protocol that incorporated many older clinical aspects, giving a pathway for diagnosis and for managing an uncertain prognosis. The temporal

ordering of the DM clinic involved subsequent trajectories of other family members, mostly younger but not exclusively so.

Latimer et al. (2006, p. 605) assert "significant advances are attributed to basic sciences and laboratory work, while major contributions arising from clinical research and practice are marginalized..." Following the advent of the DM gene test the clinic assimilated the technology into the process of reading the signs and symptoms that categorised it (Atkinson 1997). However the clinic, prior to the major discovery of the DM gene had been the site of incorporation of developments in knowledge such as linkage studies clinically, which involved "collective reasoning and adjudication" (Latimer et al. 2006, p. 604).

The DM archive documents how innovative technology such as the DM gene test emerged over time, were utilised through particular political as well as medical discourse such as the availability of the test through the NHS, and how increasing diagnostic capability does not necessarily correlate with increased "prognostic power of the physician" (Webster 2002, p. 452). The possibility that knowledge gains and innovative technology can be "generative of uncertainty and new risk" (Webster 2002, p. 453) was evident in the everyday work of the clinic in ethical issues such as testing of children and responsibility to inform the wider family of genetic risk of DM. The relatedness of practical engagement with complex issues, and sociological exploration of the culture in which advance is both made and implemented, is reflected in Webster's (2002, p. 454) observation that "the more medical technologies run through and are shaped by the social gauntlet of their construction, the more robust they are likely to be". The medical records traced how controversial issues arose through clinical practice before they were apparent in the wider context (Harper and Clarke 1990).

Relative Risk

The significance of familial beliefs about heredity has long been recognised (Blaxter and Paterson 1982; Strathern 1992a). The pre-existing way families have and use lay knowledge and beliefs about heredity is not unique to genetic conditions and constructing kinship and inheritance is part of wider discourse (Strathern 1992a, 1992b; Lupton 2003; Featherstone et al. 2006). Strathern (cited in Franklin 1995, p.

178) draws attention to the construction of kinship and awareness that "the very idea of a natural relative is a hybrid, imploded, cyborg concept, and it is a Victorian invention, not a postmodern one".

The medical records, from their external administrative changes indicating ideological viewpoints over ownership of information to their emblematic family trees, show how clinical genetics has always worked from the epistemological viewpoint of families. Notions of individuality have challenged clinical practice, especially since the advent of genetics tests such as that for DM. New genetics and the implications for incorporating healthy "at-risk" individuals into a medical framework of hereditable illness articulates a practice that is at the core of what it is to be a clinical geneticist. Atkinson et al. (2001, p. 22) describe the "simultaneous professional construction of the biological and the social" that occurs in the genetic consultation, and conclude that, in this context, "the biological is as inescapably a social product as the cultural" (p. 22).

The family tree as a symbol of kinship reconfiguration provides a graphic representation of the way an inherited condition can transform family and clinical interpretations of relatedness (Featherstone et al. 2005, p. 39). It functions as a boundary object within the clinic as it reconfigures biological and social relationships. Lindee (2005, p. 62) suggests, "...the pedigree seamlessly blends folk, emotional, social and technical knowledge into a single image and text". Kinship as a tool "between nature and culture" (Bestard 2004, p. 262) is further delineated by the construction of the DM classification as a way of contextualising the pedigree within clinical genetics.

The connection of one relative with another through potential DM diagnosis may not reflect social connection, and the DM medical records document the negotiation of family dynamics as central to clinical genetic practice. In many families the diagnosis was familiar and interviews illustrated how families constructed their own understanding of DM. This did not necessarily lead to better relationships and reflected Strathern's (1997, p. 42) observation that family as culture is context dependent and constituted out of the different interpretations of similar situations, where a solution for one person may be a problem for another.

This broader understanding facilitates insight into how people utilise genetic information and experience genetic conditions such as DM, rather than a narrow and presumptive focus on why people make their decisions (Parsons and Atkinson 1993). Through a focus on practices, in the clinical situation as documented in the medical records and the experiences related in the family interviews, the context within which decisions were made and actions explained offers potentially greater insight than an assumption that genetic information remains essentially in the same form as it travels from the clinic to the family and vice versa (Parsons and Atkinson 1993). Featherstone et al. (2006, p. 36) note how there is less emphasis on studies exploring alternative discourse of risk interpretation within families and draws attention to the potential danger of seeking "to privilege one kind of family talk or family enactment over all others, as constituting reality".

Studies from the point of view of the family experience of counselling reveal differing perceptions of inheritance, medical conditions, and risk within families (Marteau and Richards 1996; Hallowell and Richards 1997). Research indicating the potential for a negative test result to have adverse effects as well as bringing relief was apparent in the medical records (Van Riper 2005). The need for empirical evidence on which to base concepts of kin and counselling has been observed (Strathern 1992b; Parsons and Atkinson 1993; Clarke and Parsons 1997; Finkler 2005). A family tree, hand drawn by a family member and noting social achievements, contrasted with a medical pedigree within the same set of notes, reflecting Finkler's (2005, p. 1067) observation that within the context of medical genetics "family and kinship relations may be defined more by the hereditability of disorders and pain than by status and social attributes.

Previous sociological work has drawn attention to the ways genetic information is incorporated and evaluated in the decision-making processes of people in relation to inherited conditions (Edwards and Prior 2002). The tradition of Goffmann's work on stigma and spoiled identities has been applied to women's self -perception as carriers of the gene for Duchenne muscular dystrophy (Goffman 1963; Parsons 1990).

These issues raise related themes such as the genetic counselling process and the feasibility of non-directive counselling, the implications of genetic knowledge for

other family members, the responsibility for communication of genetic risk in families, and the factors that contribute to decision-making in individual situations (Hallowell 1999; Elwyn et al. 2000; Pilnick 2002; Draper and Sorell 2002; Offit et al. 2004; Loud et al. 2006). The focus of previous studies relating to the practice and evaluation of clinical genetics has primarily related to difficulties encountered by professionals in passing on genetic information to the extended family, and where responsibility for duty of care lies (Falk et al. 2003; Gaff et al. 2007).

Contemporary evaluation of the communication process between family member and genetic counsellor has recognised the need for the family context to be acknowledged rather than evaluation of the counselling process relating to professional outcomes and "unrelated to the concerns of the family member" (Clarke et al. 2005; Gaff et al. 2007, p. 999). This recognises the autonomy of decision-making whilst also recognising that technological and medical advance may seem to extend choice but that this choice may be between negative rather than positive alternatives. Feminist and disability discourse has addressed the possibility that extending choice in reproduction does not enhance liberation if it is predicated on a conformist agenda of the acceptability or not of particular genetic traits (Ettore 1999; Matthew 2005).

How individuals draw upon a range of situated knowledge that informs their genetic situation in some ways mirrors the work done in examining the public understanding of science, and the assertion that measuring the extent to which lay understanding "deviates" from experts accurately represents understanding of the issues (Wynne 1992, 1995; Michael and Carter 2001).

The way inheritance was discussed in the interviews, and was visible in the DM medical records, indicated that whereas the specifics of DM inheritance were not always clear, there was familiarity with "doing inheritance" (Richards 1997, p. 189) in terms of comparing and contrasting characteristic between family members. However the difficulties for families of negotiating different perceptions of DM may be an extra source of stress, as are issues of how to talk about DM to children and young adults (Arribas-Ayllon et al. 2007).

Summary

Sociological work immediately following DM gene discovery stressed the importance of analysing discovery in the context of the scientists' own accounts, rather than assuming a definitive story (Batchelor et al.1997). The interpretive repertoires outlined by Gilbert and Mulkay (1984) are central to this approach but analytic possibilities remain for a specific focus on the emotional investment of the scientist in the workplace. In this thesis the significance of emotions, relationships and personal meaning are explored through narrative analysis of the contemporary accounts given by the DM research team. The importance of networks and dynamics such as collaboration and competition has offered insight into the accomplishment of scientific work. The collaborative work with families DM forms an additional analytic focus, explored through the histories and trajectories in the DM medical record archive, the reconstructions of the DM scientists, and interviews with families with DM

Discourses of genetics and its definition, and disability and chronic illness, offer insight into how a condition such as DM might be experienced. Analysis of the DM family interviews draws upon this knowledge but approaches the collection and the interpretation of data from a grounded theory perspective, where emergent themes were analysed. The clinic, as a site of classification and interpretation, has formed a key aspect of sociological investigation. Its relevance in this thesis relates to the negotiation of research and service relationships between families and professionals, the impact of developing technology, and the interpretation of kinship in the context of an inherited and incurable condition.

Narrative, as a method of analysis and a way of constructing and expressing meaning, is fundamental to all aspects of this thesis. Bodily deterioration and the resulting impact on relationships and ability to accomplish self-expression led to engagement with the sociology of the body. Narrative is explored through its ability to transcend the deteriorating body through conveying aspects of personal biography no longer apparent or physically possible. Narrative analysis and the importance of retaining the context of the data allow exploration of what was said and how this was expressed. Colourful language such as metaphor has been the subject of investigation

into how concepts such as genetics are communicated and understood. In this thesis preservation of context allows close analysis of the emotional context in interviews with scientists and families. Personal meaning is interpreted as emerging from the accounts of the key actors, rather than an analytic presumption that themes will be fitted into a pre-determined structure of meaning such as an illness narrative or a story of power relations within the scientific community.

Bury (1982 p. 165) suggested a sociology "with medicine" rather than a purely social constructionist stance "of medicine". The work of sociologists such as Hallowell (1999) placed interpretation from the perspective of the person with a genetic condition centrally. Brown (1995) articulated the importance of medical sociology in influencing medical practice through communicating the experiences of people with illness or disability. Freese (2008, p. S29) acknowledges the challenges of working in the field of genetics and the discomfort felt by sociologists with biological determinism but reasserts the importance of a sociological engagement with the "social mechanisms that cause genetic differences to be more or less relevant". This thesis explores the multiple representations of DM from the perspectives of families with DM and professionals involved in the research and clinical management of a condition caused by a genetic mechanism but characterised by variability in manifestation.

Chapter Three

Characterising DM: Methods and Process

Introduction

From near the front of an unwieldy, loosely bound history of thirty years of DM in one family, a lined piece of paper with a crest printed on the top fell out. The notes were in Prof's handwriting and the contents referred to a secretor test that I hadn't seen documented in any of the other records.
I asked Prof if I could see him again informally and mentioned the test. He said he had forgotten about it but recalled how he had seen a woman, Alys, pregnant with her second child, after a chance encounter with a paediatrician led to her first child, Ben, being diagnosed with DM. Alys was the first person to be referred to the new department of medical genetics and she and her husband agreed to have the secretor test to try and establish which parent carried the DM gene.
When Prof went to the hospital pharmacy to get the solution for the test they had run out. He remembered that seeds from gorse bushes were the basis for it and described finding seeds, mixing them and carrying out the test himself. The result was non-informative scientifically and for the family but illustrates how story links details to make another world appear and how the resources for this thesis are linked and brought to life by narrative.

This thesis necessitated engagement with multiple methodological possibilities in order to fully explore themes emerging from data sources varying from documents to voices. Analysis of the language and emotions of key actors facilitated insight into the process of landmark scientific discovery and the consequences of this for professionals and families. Characterising refers to the scientific description of defining a particular gene and to narrative and the insight that characters bring to experiences and events.

Resources

Outline

The research involved documentary analysis of forty- two DM medical records, semi-structured interviews with twenty people with a DM diagnosis from eighteen families, and semi-structured interviews with six members of the DM gene discovery team. Access to these three different data sources required distinct approaches in terms of ethical approval, access and analysis.

The DM archive and register were located in the Institute. The DM register was accessed to recruit family members for interview. Of the six professionals interviewed one was a laboratory technician, one was the lead clinical geneticist for the Institute, and four were laboratory scientists at varying degrees of seniority in

their careers. At the time of the current study none were employed by the NHS, two were still working at the Institute, three were at other universities within the UK and one was working abroad. Prof had retired and was working as an emeritus academic researcher associated with the Institute.

Although the scientists may be recognisable by inference their contributions have been anonymised and they have been given pseudonyms, detailed in Chapter One.

The DM archive and DM register

The medical record is a fundamental aspect of medical work and as such represents an important focus for sociological enquiry (Berg 1996; Berg and Bowker 1996; Hobbs 2003). According to Hobbs (2003, p. 473) "A comprehensive understanding of medical discourse requires the study of both oral and written communications", reflecting previous observation on the significance of documentary research and the relative lack of work in this area (Atkinson 1995; Hammersley and Atkinson 1995; Prior 2003). The DM archive is part of the everyday work of the Institute. The file numbers and sequence reflected the efforts over time years to organise the records and respond to increasing knowledge, which in turn impacted on everyday practice, both bureaucratic and medical. The majority of the records were in three main sequences of numbers and located in two locked cupboards. They were located in a building requiring intercom access for non-permanent staff that is separate but close to the Institute. Institute staff, including the two geneticists who run the muscle clinic and their support staff, occupy one floor of the building.

The DM research register was established in 2004 in the Institute, and funded by the Myotonic Dystrophy Support Group (MDSG) to maintain an up-to-date database of family members interested in taking part in research projects. It was registered with the local NHS database protection officer and has a strict protocol for access and use. To date it has been accessed for local projects but it is possible that it could be used in conjunction with other muscle centres for clinical trials. It is designed for use with the DM clinic protocol. The geneticist should fill the standardised form every time a patient attends a DM clinic run by the Institute. Appointments are usually annual although they can be given on request if necessary and are held in the hospital

outpatient department. All medical records and any necessary equipment necessary are taken to and from the clinic.

The researcher

The project was grounded in "methodological self-consciousness and a concern for reflexivity" (Van Maanen 1995 pp. 7-8). Researchers bring experiences, values and attitudes that may not be immediately obvious, even to them. They contribute to the production of the interview, "operate from within a theoretical overview...and affect the data at all stages" (Scott 1985, p. 74). Previous familiarity with the research setting meant that some insight could potentially be lost. The researcher, who knows the environment through engaging in it in another role, may not at first see the unfamiliar in the familiar.

I had previously trained as an occupational therapist and been employed as family care officer by the Muscular Dystrophy Group in the 1990's and as a research officer by the MDSG in 2003. The work was based in the Institute, although its emphasis was on providing emotional and practical support, rather than genetic counselling, and meant that there was familiarity with aspects of muscle disease. The roles involved attending the specialist muscle clinic, carrying out home visits and setting up a research register.

Taking on the role of researcher involved "making the familiar strange" (Delamont and Atkinson 1995, p. 3). The rationale for visiting families, interviewing scientists or accessing the medical records from a sociological perspective was different to that of a support worker. Adopting this perspective prior to data collection was challenging but the real learning came about through being in family homes.

There is also the question of how much the presence of the researcher affects the research process (Hammersley and Atkinson 1995). This has been the subject of ongoing debate from posititivist ideas of collecting a "true" picture of reality to the post-modern and feminist approaches, which acknowledge the presence of the researcher as co-constructing the text and of contributing to the research process at all stages (Borbasi 2005). The feminist standpoint advocates a fully engaged

reflexive position characterized by "intimacy, self-disclosure, reciprocity and caring" (Tong 1995, cited in Borbasi 2005, p. 498).

The choice of qualitative, semi-structured interviews was taken to maximise openness but reflexivity at every stage of the interviews led to a conscious engagement of the reality of interviewing as "conversation with a purpose" (Burgess 1984, p. 102). Denzin and Lincoln (2000)) state that the interview is a co-constructed act. The methodology was chosen for its suitability in "allowing respondents to be seen as individuals with social histories and unique perspectives on the world" (Borbasi 2005, p. 10). There was no expectation of a single narrative of DM and the focus was on how DM is constructed by a multiplicity of perspectives and experiences.

According to Lipson (1989, p. 65)) optimal data collection comes from "relationships in which informants trust the researcher and in which the researcher has a grasp of his or her own influence on the interaction". Therapeutic purpose was never actively suggested or pursued in the course of the research. However in many interviews the families spoke of the benefits of talking about DM with a person associated with it. The participant information sheets gave details of clinical contacts should the interviews prove upsetting.

Research design

Epistemology

Qualitative research accommodates the complexity of the social world and its methods allow the exception as well as the rule. There is a spectrum of methods from which the most appropriate are chosen based on the research aims (Flick et al. 2004). The varied sources of data necessitated close engagement with methods and ongoing appraisal of their optimal use.

The interpretive paradigm within which the methodology for this project is located is constructivist grounded theory (Denzin and Lincoln 2000). I was aware of the criticisms of relativism but took the approach of Lupton (2003), where the main focus of the research was a close examination of the work that was being done to

make the setting of genetic medicine, the definition of DM and the experiences of professionals and family members visible.

Documents as research topics

The DM medical records were treated according to Scott's definition of a document "...as an artefact which has as its central feature an inscribed text" (1990, p. 5).
Scott (1990, p. 14) refers to archival materials as being "open to all comers...subject only to minimal administrative restrictions" but in this context the DM archive comprised a physical setting where documents of both historical and contemporary significance were housed. Access to them was limited and stringent, but this assisted in the establishing the authenticity and genuineness of the documents, both key considerations in using documents for research (Scott 1990).

The analysis of any document is dependent on literal and interpretive meaning (Scott 1990, p. 29). In the DM medical records the literal meaning was encoded in the language of genetics, so documentary analysis entailed becoming familiar with this vocabulary. There was no disputing literal meaning as it reflected contemporary practice and can be validated in the present, rather than relying on historical clarification. However assessing the interpretive meaning required a number of different strategies and approaches, including looking at the medical records in terms of content and context in order to orientate them within the organisational culture in which they operate, and the wider social setting to which they relate. They were studied "as socially situated products" (Scott 1990, p. 34). Documents do not exist in isolation, and are governed by rules and conventions, which are necessary to engage with in order to appreciate their social significance (Scott 1990; Prior 2003; Atkinson and Coffey 2004). MacDonald (2001) notes that in addition to being social products, documents are socially produced. May (2001, p. 183) observed that documents "do not simply reflect, but also construct social reality and versions of it".

Research design acknowledged the key area of enquiry that locates the document within a particular context. The DM medical record was governed by external requirements that it fulfil the criteria of medical notes. It also reflected a developing bureaucratic identity as the field of medical genetics expanded and formalised. It contained traces of the configuration and reconfiguration of DM from the different

perspectives of the writers and the scientific progress conveyed by the laboratory test results.

There was recognition that "interpretive meaning ...is ...a tentative and provisional judgement which must be constantly in need of revision..." (Scott 1990, p. 35). As a researcher I was not immediately privy to the world of taken-for-granted knowledge in which the medical record was produced. Despite previous familiarity with the general setting, I had little knowledge about the way the notes had come into being. Their meaning was embedded in practice and this differed according to who was handling them. Although the geneticist primarily uses the DM records, there were also traces of bureaucratic development as the institution itself began to emerge from a small specialist department to an independent regional centre. A pilot study followed by in-depth investigation of the medical records allowed multiple voices to emerge and brought the documents into the context of the everyday world in which they operate.

Document analysis was not seen as representative of institutional practice (Coffey and Atkinson 1996). Documents operate within their own context or network and were used in conjunction with semi-structured interviews to gain a deeper understanding (Scott 1990; Atkinson and Coffey 1997; Prior 2003). The aim was to investigate how the visible traces on the pages were the end-product of invisible processes governing their existence as "receptacles of inert content", but also as "functioning agents" (Prior 2008, p. 821, p. 830) in their own right. Documentary analysis was located in the wider research question of how the scientific progress, traced in the notes, influenced everyday practice and experience. This was broken down into research questions that addressed: the changing definitions of DM over time, the experience of being involved in scientific discovery rather than the accounts in scientific journals, the relationship between families with DM and researchers, the nature of service and research in institutional development, and the everyday experience of living with a condition which has mostly been described in the words of professionals. The observation that "Without documents there are no traces. Things remain invisible and events remain unrecorded" (Prior 2004b, p. 375) was further interpreted to note that even within documents selected events could be made more visible than others.

Families and family members were represented in the medical record through the prism of inherited disease and they appeared in their own words in letters written to Institute staff, but their experience in the medical record was overwhelmingly conveyed in the words of professionals.

Of primary interest was the place of the medical record in a context of uncertainty, development and change where issues initially grounded in scientific advance rippled outwards to incorporate wider social and ethical issues related to genetics as a new way of interpreting the body.

Narrative

Locating the research context involved reading the history of narrative within sociology and narrative analysis as a method, including the ongoing discussion as to how it is best interpreted. Labov (1997, p. 395) notes:

The discussion of narrative and other speech events at the discourse level rarely allows us to prove anything. It is essentially a hermeneutic study, where continual engagement with the discourse as it was delivered gains entrance to the perspective of the speaker and the audience, tracing the transfer of information and experience in a way that deepens our own understandings of what language and social life are all about.

Storytelling is ubiquitous and also local, in its conventions and norms. Mc Call and Wittner (cited in Denzin 1990, p. 59) describe how ordinary people "create culture" when they tell stories. Through accessing the medium of storytelling the narrator communicates experience and meaning that may or may not be familiar to the hearer. Culturally accessible tropes such as metaphors allow the unfamiliar to be translated into more meaningful concepts for the audience. Although distinctions can be made between using the words narrative and story a distinction was not made in this thesis (Riessman 1993; Lieblich et al.1998; Gergen 2001).

Hart (1998 p. 8) writes of the necessity for integration in academic work and defines it as "making connections between ideas, theories and experience" and characterised by "systematic questioning, inquiring and a scrutinizing attitude". According to

Denzin (1989, p. 27) "The word *method* shall be understood to refer to a way of knowing about the world". Riessman (1993) writes of the search for a method in the 1980's and how traditional qualitative methodology did not fit with the narrative sequences in transcripts for analysis. However, guided by Mishler, Riessman (1993, p.vii) found a way of interpreting the narratives through "close textual analysis: seeing how a narrative is constructed and how a teller rhetorically creates it to make particular points". This places the study of narrative in the "interpretive turn" within the social sciences (Geertz 1973; Riessman 1993). Denzin (1989, p. 28) describes how "*Interpretation*...creates the conditions for *understanding,* which involves being able to grasp the meanings of an interpreted experience for another individual".

The focus of interest is the story itself. Within the history of social science this was a move away from a research model adapted from the natural sciences (Bruner 1990; Sarbin 1986). The origins for more qualitatively based methods are found in the Chicago School in the 1960s. Ochs (1997, p. 185) traces the literary preoccupation with deconstructing narrative to the 1962 translation and publication of Aristotle's "Poetics", stating that since then "narrative genres such as tragedy and comedy have been the preoccupation of philosophers and critics". However up to the 1980s authors such as Riessman and Polkinghorne were arguing for the place of narrative within sociological research. Both came from practitioner backgrounds and recognised the significance of narrative in therapeutic practice. They argued for the extension of narrative into research. Their work was influenced by the literary theory of writers such as Barthes (1966) who were concerned with the structure and functions of narrative. These functions were personal, allowing an individual to construct a self-identity through narrative, and cultural allowing the expression and transmission of shared values and beliefs.

The analysis of narrative presents an epistemological challenge as "the form of that analysis depends largely on the researcher's views on the construction of knowledge" (Redwood 1999, p. 674). Phillips (1994) notes that utilising data from narrative has been questioned. The central issue is the concept of the "truth" of the narrative. There are many ways of interpreting the data which will be discussed but before that there is the challenge of deciding what is expected of the data. Is it an

actual representation of the experience and meaning of the narrator that can be "captured" as a definitive text, or is it a subjective account, which depends for its construction on the context in which it is being formed as well as the experiences in which it is grounded? Goffman (1981 p. 504) drew attention to the fact that "The apparent vivid spontaneity of performance should not cause an analyst to lose sight of the prefabricated" while Mishler (1986) wrote that the coherence of accounts does not necessarily make them true. The epistemological basis for this thesis acknowledges that the context in which a narrative is constructed is relevant to its interpretation.

In their introduction to a methodology for narrative research Lieblich et al. (1998 p. 1) note how this objective may seem to be in contrast with the narrative approach itself which is more easily aligned with literature with its basis "on talent, intuition or clinical experience" and seeming defiance of order. However they argue that divergence from a positivist approach, with its assumption of a single truth or interpretation of a text, to a more subjective and relativist epistemological position does not preclude an exposition of method (Lieblich et al. 1998). They examine the universal use of narratives and their significance to personal and social identity.
Gergen (2001) explores the nature of truth from a different perspective when he discusses how narratives are replete with characters that are necessary to illustrate the narrator's perspective. However he notes that these characters must uphold the narrator's point of view otherwise they present a challenge to the narrator's identity. Identity therefore is reciprocal and "In most instances the actions of others contribute vitally to the events linked in narrative sequence" (2001, p. 258). This suggests that accounts have an inbuilt capacity for integrity in that they are available to be validated by anyone and are not the property of the narrator. This does not mean that a particular story is actually "correct" in every factual detail but rather that there is a repertoire of functions which the story must fulfil in order to be plausible. Gergen's (2001, p. 258) observation that "a fundamental aspect of social life is the *network of reciprocating identities"* is meaningful to the analysis of the data for this thesis. It resonates with the analytic proposition that the scientific team reconstituted themselves retrospectively as meaningful and important to each other in their memories of the time of gene discovery. This narrative enactment of the

work that had gone into gene discovery was given emotional recognition in their individual accounts of what was significant to them about the time.

The position of the interviewer is open to consideration when a story is located within the context of an interview, as in the case of this thesis. Gergen (2001, p. 257) writes, "Narration may appear to be monologic, but its success in establishing identity will inevitably rely on dialogue". The interviewer has dual functions as they are allowed ask the questions but they are also the audience. Mishler (1986, p. vii), while accepting the importance of interviewing as a research method, challenged the way interviews were interpreted and offered the view that "the interview is a form of discourse".

The influence of the researcher in analysis, as well as the recognition that they are not a neutral presence in the interview itself, is also the subject of debate. According to Riessman (1993, p. 22) "Narratives are interpretive, and, in turn, require interpretation". The initial transcription of the text, with decisions about the level of detail and non-verbal events to include, is a theoretical one that has later bearing on analysis. The way an utterance is made may influence its interpretation by an audience. The importance of always going back to the original text is emphasized as the way of obtaining analytic validity. The theoretical basis for analysis overlaps to some extent with the epistemological approach of the analyst to the issue of truth. Stivers (1993, p. 424) writes that interpretative analysis aims for "believability, not certitude, for enlargement of understanding rather than control".

There is considerable overlap between psychology and sociology in the recognition of the fundamental importance of stories as ways to "provide coherence and continuity to one's experience" (Lieblich et al. 1998, p. 7). Phenomenologists such as Polkinghorne, and personologists such as McAdams, while viewing the nature of knowledge differently, all attach significance to the individual agency of the narrator (cited in Gergen 2001). Bruner (1986, 1990) subscribes to the importance of agency and the self but attaches equal importance to the cultural context in which the story is constructed. However they are all in agreement that "The story *is* one's identity, a story created, told, revised and retold throughout life" (Lieblich et al. 1998, p. 7). Stories contain essential events and the circumstances in which they

occur, but they are also formed as they are told (Holstein and Gubrium 2000). The performative aspect of a story to some degree influences its survival in the social world. Use of devices such as metaphors and other literary tropes assist in engaging the audience (Coffey and Atkinson 1996). These are based on local conventions and thus the events related in a story may receive greater or lesser emphasis depending on the context in which it is told.

Recognition of the presence of the interviewer leads to the question of the "self" that is being narrated. For Goffman (cited in Collins 1988, p. 49) the self was not a private individual but "a socialized entity, created in and through social interaction". The narrative is the vehicle for self-expression that is performed and governed by organizational principles called frames. These frames refer to the way there may be several subjective realities operating simultaneously during a performance.

Goffman's theory allows for the presence of the audience and its ongoing evaluation of the performance to be recognised. The viewpoint taken in this thesis is akin to Bruner's acknowledgement of the influence of the cultural, social context but also the existence of the self and internal agency.

Memory

Memory was recognised as integral to the gathering and interpretation of the data. It was closely related to the approach of narrative analysis used to interpret the interview accounts. Aspects of memory were essential to a reading of the analysis, such as the relationship of memory to meaning and the relationship of the individual to the culture of which they belong. The iterative process of attributing meaning to the past through repeated interactions places it within a social framework (Halbwachs 2001, cited in Jedlowski p. 31).

Narration is the medium through which this meaning is accomplished and repeated. The way memory operates at different levels of interpretation is similar to the way narrative analysis takes into consideration not only what is said but how, and the social context in which it occurs. The relationship of memory to identity acknowledges the many possibilities available for the individual to select their

account, and also how memory serves to shape and transmit a specific cultural as well as individual identity.

Narrative analysis of the scientific interviews revealed common themes of occupational identity and culture, shared meanings about the practice and process of science but also important differences in the personal meaning of the gene discovery. This highlighted the work of memory in action, as events from the past were infused with emotion and language to convey meaning, both individual and collective, to a contemporary audience.

Scientific definition rather than scientific culture is represented in the publications of the scientific world. The process of reduction means that contingencies and problems do not appear and "scientists themselves write such agency out of the narratives they publish" (Gooding 1992, p. 66). The potential for narrative to "reveal multiple truths and more evocative, revelatory pathways for dialogue and understanding" to challenge the reductive approach of scientific or medical definition where "fragmented, messy lives are reduced to information" (Alexandra 2008, p. 101) applied to both the scientific and family narratives. Through their narrative accounts constructing the personal meaning of the DM gene discovery there was the potential to allow other scientific genres other than the "narrative leviathans" (Traweek 1992, p. 442) endlessly portraying cause and effect.

Interviewing

DM is documented in medical literature and the DM gene discovery in scientific literature but this thesis is concerned with everyday experience and practice. Qualitative interviewing was interpreted as a method of uncovering and exploring the meanings that are integral to people's lives and allow "us to see that which is not ordinarily on view and examine that which is looked at but seldom seen" (Rubin and Rubin 2005, p. vii). The viewpoint of the study was that "Qualitative interviews examine the context of thought, feeling and action and can be a way of exploring relationships between different aspects of a situation" (Arksey and Knight 1999, p. 32). According to Sarantokos (1998, p. 58) "Methodologies are the tools of the trade of social scientists" and within the choice of qualitative interviews as part of the research design there were choices about interview type.

The interviews were seen as "active" (Holstein and Gubrium 2000, p. 141), recognising their interactional and interpretive nature. Qualitative methodology implies that the discourse is situated and constructed by the interviewer and the interviewee (May 2001). It recognises the role of the interviewer as being part of the process of meaning making (Briggs 1986; Silverman 2001). This had practical implications, in terms of prior acquaintance with the scientists and knowledge of the everyday setting of their work. As this knowledge did not relate to scientific expertise it was necessary to prepare and become familiar with the sequence of events as they occurred in the scientific community.

An essential part of being an interviewer is the ability to listen, as well as to know when to ask a question (Mason 2002). This refers to the necessity to give due attention to the respondent as part of the process of establishing rapport, but also to the ability to listen for areas of particular interest which could be probed more deeply.

Literature on fieldwork can imply that it is a series of orderly and predictable steps whereas reality shows that it is a mixture of practical and mental preparation and dealing with contingencies (Borbasi et al. 2005). In deciding on research design the question of the skills of the researcher are important. Qualitative interviewing "belies a much more rigorous set of activities" (Mason 2002, p. 63), including preparation and planning, development of communication and listening skills, and analysing the collected data. Within the choice of interviews as a method there is a wide range and May (2001, p. 121) stresses the importance of the researcher closely examining these in order "to understand the different methods of conducting interviews and analysing the data, together with an awareness of their strengths and limitations". Arksey and Knight (1999, p. 3) also stress that interviewing comprises a set of methods and that choices of what type of interview to use "are complex decisions that shape the potential meanings of our findings". When further refining the interview to a particular type care should be taken that the choice "is aligned with the strategy, purposes and research questions" of the project (Punch 2005, p. 170). For example, within the choice of semi-structured interviewing it was decided to use the life story method (Atkinson 1988) with Prof, as his career was closely bound with progress in knowledge about DM and the development of the Institute. The ontological position

of the interviewer in using the qualitative interviewing approach is one that "values people's knowledge, values and experiences as meaningful and worthy of exploration" (Seale 2004, p. 182).

A voice recorder was used for all of the interviews. There are three areas of consideration in using the recorder: "interaction, transcription and interpretation" (May 2001, p. 137). The professionals were familiar with interview technology but family members were less likely to be. However using it enabled me to concentrate on the respondent and build up rapport, rather than taking notes, which may have evoked medical appointments.

The semi-structured interview

The choice of semi-structured interview was made to allow some structure but also allow scope for rich description and to "follow up ideas, probe responses and ask for clarification or further elaboration" (Arksey and Knight 1999, p. 7). Foddy (1993, p. 25) stresses the need for the interviewer to clearly define the topic, the respondent to have the information necessary to answer the question and the respondent's ability to verbalise the answer in the interview situation. The interviewer and the respondent are in a constant state of encoding and decoding each others questions and answers and are engaged actively in meaning making (Foddy 1993). Semi- structured interviews allow the interviewer to adapt to "the level of comprehension and articulacy of the respondent" (Fielding and Thomas 2001, p. 124).

Atkinson (1998, p. 40) recommends an "informal approach, eliciting open-ended responses and in-depth comments" and Spradley (1979, pp. 86-88) advocates the "grand tour" question as a way of opening up the interview and allowing the gradual emergence of themes. The use of open questions allowed the complexity of these themes to be explored. The focus of the interview was to listen to "informant's accounts of their behaviours, beliefs, feelings and actions" (Arksey and Knight 1999, p. 4).

A loosely structured interview schedule was prepared in advance of the interviews (Appendix one). Its main purpose was to introduce the areas of interest. The initial question was intended to open the interview and allow the respondent the

opportunity to speak about an area that was familiar and non-threatening. Each interview finished with a question inviting the respondents to talk about anything they felt had not been adequately covered during the interview.

The life story interview

In choosing a life story interview the researcher's epistemological position is one that recognises the inductive, theory building nature of qualitative research. According to May (2001, p. 134) this type of interview is akin to "detailed conversations which attempt to give a fuller insight into a person's biography". The aim of this interview was to explore how scientific discovery was experienced by a professional whose role was both scientific and clinical, and whose career spanned major advances in DM research. Prof was considered an "elite" as he has achieved international recognition within his profession. Gene discovery is now familiar and the aim was to explore this taken-for-granted knowledge and the context in which a young doctor became interested in a then relatively unexplored field of work. Rather than looking at the work itself the interview investigated how the work was achieved, the ways in which Prof made sense of his career, and his interaction with the world of scientific discovery. This has been described as "documenting the inner experience of individuals, how they interpret, understand and define the world around them" (Faraday and Plummer 1979, p. 776).

Prof was accustomed to the interview situation and the challenge lay in "helping the person create and convey his or her meaning in life through the story of what has happened" (Atkinson 1998, p. 40). The focus of the interview was to listen to "informant's accounts of their behaviours, beliefs, feelings and actions" (Arksey and Knight 1999, p. 4). The life history interview is relatively unstructured but this does not mean it is ill defined. The choice of a method giving scope for emergent themes was justified as Prof was motivated by the project itself and was in a unique position to give an overview of a time of rapid scientific change, which impacted on society in terms of moral and ethical issues such as genetic testing.

The email interview

The reason for choosing online interview for one scientist, Bill, was geographical distance. Issues such as choosing participants, designing the project, and ethics are

similar in both Internet and traditional research, but an issue unique to the Internet "is the potential difference between synchronous and asynchronous communication" (Hewson et al. 2003). Email is less immediate but more engaging than asynchronous communication (Mann and Stewart 2000). Response to initial contact about the project was positive. Following email discussion it was jointly decided that asynchronous communication would facilitate ease of communication and convenience for Bill. It also allowed more time to reflect and respond to the questions. Asynchronous e-mail has been described as the least interactive type of computer-mediated communication (CMC) but also as being akin to letter writing where there is time to reflect on the questions and responses (Mann and Stewart 2000)

The use of the online interview reflects the speed of technical advance in a contemporary culture sometimes negatively referred to as an "interview society" (Silverman 2004, p. 22). There is continual updating of information on the efficacy and etiquette of using computers as a research resource (Mann and Stewart 2000). The phrase "netiquette" describes the "established conventions" and "standards of being social and relating in the online environment" (Mann and Stewart 2000, p. 14). The validity of online research as a research tool has been debated and there were refinements to be made before communication was established. Issues such as informed consent, a clear understanding of what the research interview entailed, security and confidentiality were equally important whether using face to face (FTF) contact or CMC. The familiarity of the interviewer and the respondent with the technology is an important aspect as there are conventions that are specific to online communication such as emoticons and abbreviations (Mann and Stewart 2000; Seale et al. 2004). The question of whether CMC is appropriate has been the subject of debate with negative perspectives focusing on worries relating to empathy and richness of data (Kiesler and Sproule1992; Smith-Stoner 1999). However there is also a view that people can express their personalities as addition to their opinions in a wide range of technological environments (Walther 1992). There are issues related to this method such as the lack of non-verbal cues and para-linguistics but "it is down to the individual researcher to decide whether an Internet methodology will provide a better alternative to more traditional interview techniques" (Hewson et al. 2003, p. 45). The ability to think carefully about responses, particularly as the events

themselves occurred many years before, was considered to be a more important factor than the loss of spontaneity associated with a traditional interview.

It was important to bear in mind the fact that CMC is "text-based, contextual information" (Johns et al. 2004, p. 244) which cannot convey the subtleties of intonation, facial expression or gestures. The text has to be crafted so that it can stand alone without the presence of the interviewer to clarify any misunderstandings or ambiguities.

Research protocol

Ethical considerations were a factor in the research design and process as "ethical conduct provides the basis which legitimates the whole enterprise" (Payne and Payne 2004, p. 66). The choice of methods gives indications of issues that may arise, but the process of data collection may result in unforeseen circumstances where response to an ethical issue is required. Mason (2002, p. 7) describes conducting qualitative research as "*moral practice*" where the researcher demonstrates "sensitivity to the changing contexts and situations in which the research takes place.

The model used was the research protocol. This is "an official account of the intended research methods and procedures, with special attention to how benefit is maximized and risk minimized, autonomy is respected and fairness to subjects is ensured" (Sieber 1992, p. 14). The advantage of looking at ethical issues within the framework of a research protocol was that it was developed at the start of the research planning and incorporated into the research process. Although it is not possible to anticipate every situation, it formalised the place of ethics within the project. The protocol included prototypes of the letters and information packs sent to respondents and an outline of where and how the data was be stored, recorded and analysed (Appendix two).

The choice of research topic was critically assessed for its potential relevance, as data collection is dependent on the goodwill of participants who expect a rigorous approach to both research design and process (Denscombe 2002).

Ethics

The guiding principle was that the project aims were understood and ethically approved before data collection. Ethical approval involved two separate applications. The DM research register and archive were part of the NHS regulatory system.

The medical record is a legal and medical document and there were ethical issues associated with accessing it for research purposes. The framework for accessing records or recruiting families requires compliance with NHS regulations. Application was made through the NHS research and development (R & D) committee of the hospital where the records were located, and then applying to the local research and ethics committee (LREC) using a standard computerised NHS COREC form. Receipt of the form and a date for its hearing was communicated by the REC by email and post. The medical supervisor based in the Institute attended the hearing with me. The ethics approval reference is 06/WSEO2/116.

Ethical approval for interviewing Prof and the scientists was sought through the School of Social Sciences Research Ethics Committee using the standardised procedure, which fulfilled the criteria for approaching respondents employed by academic institutions or private companies. Approval was given without delay or amendments.

Hewitt (2007, p. 1150) addresses ethics surrounding interviews that elicit pain or distress and the unquestioning use of qualitative interviewing as a research technique, which may result in anecdotal or a "suspiciously cohesive summary of responses". Accordingly "A moral obligation then exists for the researcher to ensure that there is sound justification for the investigation and the research method" (Hewittt 2007, p. 1150). All families were approached according to the research protocol and received an information sheet outlining all aspects of the research. The stringent guidelines of the ethical committee included references to confidentiality, and protection from harm. However the nature of interviews meant that it was not possible to foresee every potentially upsetting circumstance. Hewitt (2007, p. 1153) lays down practical guidelines relating to autonomy, vulnerability, beneficence and justice. In practice before each interview began its purpose was discussed again especially in view of previous involvement with families with muscle disease. Informed consent was obtained at each interview to protect autonomy. Inherent in the

choice of method was the viewpoint that research analyses are versions of reality. The aim was to base research findings on a foundation that would not distort or generalize the actual experiences and words of the family members through assuming their intention or format. The balance between causing upset and acquiring data lay in the validity of the research question and the recognition that talking about living with DM may be upsetting.

The similarities between therapeutic and research interviews, and the potential to cause harm or benefit have been noted (Stacey 1988; Merrell and Williams 1995). Goodwin et al. (2003) draw particular attention to the complex relationship between rapport and exploitation and argue that where rapport is greatest there is also greatest danger of crossing boundaries.

The potential for distress to the researcher is an important facet of reflexivity (Coffey 1999). The research contract with the families was based on stating the research purpose so as not to exploit boundaries based on previous work and experience in muscle disease. The research protocol provided guidelines but the contingent nature of relationships entailed using judgement. Leaving a person who had become upset in the interview was difficult but their experience was valid and essential and my role as researcher was to explore this without exploitation.

Confidentiality is integral to the ethical procedure, and in cases where anonymity was difficult to protect, such as the publicised accounts of gene discovery, this was acknowledged (Hammersley and Atkinson1995). Although respondents were anonymised there was awareness that the general setting and sequence of events could be recognised. All people referred to during the course of the interviews were also anonymised but place names were not. There was recognition of the fact that there are many published papers of the gene discoveries already in the public domain and complete anonymity is difficult. However where there was the possibility of harm, such as potential suicide being discussed in one family interview, the family member was asked to allow me to contact the DM geneticist at the Institute.

The information sheet given to each respondent contained details on data collection and storage in compliance with Data Protection requirements and clearly stated that there was no obligation to take part in the research

Recruitment

The protocol for recruiting family members from the DM register necessitated registration of the project and agreement to provide feedback to the MDSG at the end of the project.

The senior DM geneticist at the Institute identified thirty- eight out of fifty- five people on the register to be contacted. Exclusion criteria were changes in circumstances that made contact inappropriate such as illness. Introductory letters and information packs were sent by post and potential participants were contacted by phone approximately one week later. Twenty- three people responded positively and it was possible to arrange twenty interviews. Interviews were arranged at a time and place of the family member's choosing. In all cases this was the home.

As I had previously worked in the same department as the gene discovery team an initial email contact was considered appropriate to ask whether a letter of introduction and information pack could be sent. The response was positive and information was forwarded by post or email. A time lapse of one week was allowed, as indicated in the information pack, before being contacted again by me. In five cases the scientist replied immediately to say they would like to take part and in one case subsequent to the second contact being made. There followed further email and telephone contact, establishing suitable times and venues for the interviews.

The Real World

Research design shaped and was shaped by the research process. A grounded theory approach meant that themes arose out of the data rather than from a pre-defined framework. It influenced how the data was gathered and was an exciting, iterative process that demanded reflexivity and constant evaluation. Every aspect of the project, from getting ethical approval to accessing the DM archive involved dealing with people. These dynamics determined the course of the project as much as the intellectual framework.

Home visits

Twenty people in eighteen families with DM were interviewed. Two interviews involved two generations of the same family, and in nine interviews other family members, generally partners, joined in. The interview participants ranged in age from early twenties to mid-seventies. The interview locations ranged from city to more rural areas. Although it was possible to recognise the features of myotonia and the associated tiredness in individuals, this was not a homogenous group in terms of how DM manifested. This concurred with the research aim of being open to the spectrum of experiences associated with DM.

There was awareness that every situation was new and different. Interviews where more family members joined in were unexpected and the first time it happened necessitated thinking on the spot about how this might affect the research. The deciding factor in every interview was the context of being a guest in a person's house and respecting their boundaries. The REC approved protocol was followed so that by the time I arrived the family member had all of the project information and then gave signed consent.

Spradley (1979) refers to the first five minutes of any interview as being crucial in establishing rapport. A ritual associated with welcome is offering a cup of tea, and in most cases this occurred. Acknowledging this convention, while judging when it was appropriate to accept or not, was important as in several instances the person was extremely frail.

The degenerative nature of DM meant that speech was often slurred and quiet. It was necessary to place the voice recorder as close as possible to the person. The interview transcripts reveal the extent to which speech can be affected. One interview could not be understood by the professional transcription service. Although difficult to understand I found it was possible to extend the transcript considerably by multiple listenings. This raises questions about familiarity with altered speech or speech difficulties when transcribing interviews. The interviews often involved asking people to repeat what they had said and judging when that effort would inhibit the flow of conversation. Although some loss of data is acknowledged, every interview

contained codes that later became integrated into the key themes. The interviews, which were professionally transcribed and then re-checked by me, also revealed gaps in the interviewer questions where the question was not fully formed. In some instances this appeared to be due to my accent, but it rarely interfered with the responses. This parallels the feminist perspective on the research relationship where "the specific wording of a question is less important than the interviewer's emotional attentiveness and engagement and the degree of reciprocity in the conversation" (Riessman 2008, p. 24).

Listening is an active rather than passive behaviour. Preparation for interviewing involved reflecting on listening skills and breaking them down as an activity, considering non-verbal behaviours like eye contact and posture, and reflecting skills such as paraphrasing.

Reflexivity was key to establishing rapport in the interview relationship. Fredriksson (1999, p. 1167) distinguished between two conversational modes of nurse- patient interaction: relating "being there" to caring contact whilst carrying out a task, and "being with" to non-task related listening. This distinction provided a useful way of considering relationships. My previous familiarity with home visits focused around practical help, and this led to initial feelings of helplessness and guilt. However by actively listening and clearly outlining the purpose of the research it was possible to concentrate on the families. This resulted in less reliance on the aide memoire and a more instinctive approach. Focus before the first interviews was on practical preparation but this changed to one of mental preparation to allow for full concentration on the act of interviewing, rather than the framework surrounding it. This resulted in an active effort to shed pre-conceptions.

The participant information sheet referred to patients and gene discovery. The experience, from the first interview, of being welcomed into a home and having a conversation where the person communicated their experience of DM through the framework of everyday life rather than genetic disease, led to the first changes. The term "patients" did not relate to a person living with a chronic incurable condition outside the medical context. Asking directly about gene discovery did not open up the interview but led to general research related responses. However from early in

the process I felt that this need not inhibit the research and if the research interview became even less structured there was the possibility of the impact of research progress emerging from the family perspective. As Riessman (2008, p. 24) noted "Creating possibilities in research interviews for extended narration requires investigators to give up control, which can generate anxiety" but this was felt intuitively to be the best way to allow the experiences of the person to emerge rather than try and impose categories of experience by rigid questioning. This does not imply that the questions asked were random and unconnected to DM as they generally started with asking the participant how they had become aware of DM. In this way a picture of DM, with common themes across all the interviews, began to emerge but one that I hoped recognised the agency and expertise of the person with DM. The iterative process of carrying out the interviews reinforced my initial view that if gene discovery was significant to families it would emerge through their accounts of how DM influenced their lives. The decision to place little structure around the family interviews necessitated listening and an intuitive approach that the necessary material would come out of the interview, rather than it being dominated by a limited range of enquiry.

The way a research interview is an interpretation rather than a carbon copy of the protocol, was apparent throughout. In one home a child came into the living room, and without advance discussion his parents changed the topic immediately, telling him that I was a financial advisor. They explained later that he had not been told of his mother's diagnosis or the possibility that he might have DM and that this was the first time that they had discussed this together. In another interview a young man spoke repeatedly about suicide, moving between accounts of despair to accounts of coping. He was alone and as the interview progressed the focus shifted from a research interview to analysing the encounter as it was happening. Respecting his agency provided a means of talking about getting the help he felt was most appropriate for him through the DM clinic.

Conversely being familiar with the manifestations of DM meant they were not a worry or a distraction. There were positive aspects of familiarity such as experiencing no difficulty in managing time lapses in conversation due to muscle weakness. Witnessing the many ways DM impacted on people reinforced its

textbook classification as a variable condition. Some of the people interviewed showed little visible signs of the condition. In other instances people crawled around their living room or sat in a wheelchair. Drooping of the eye muscles due to weakness meant that in one interview I didn't know if the woman was crying at times.

A seemingly simple aspect of listening such as letting the speaker finish is important in a condition like DM where there may be a delayed physical response. Consideration of the physical implications of DM, and knowledge of manifestations such as excessive daytime sleepiness, lack of facial expression and possible delay in responses were essential to the pacing of the interviews (Harper 2001). Tiredness is a key feature of DM and the agreed interview time was no more than one hour. In some cases the person was visibly tired before this time. In interviews where the person wanted to continue the time was acknowledged and the choice left with them about continuing.

Listening has been described as a most effective therapeutic technique and although this was not the purpose of the research several people alluded to their sense of wanting to talk and of talking being helpful. An important aspect of beginning and ending the interview was to place it in context and to refer to further help if it was necessary. Several of the family members became upset and cried and it was important not to leave until the person had the opportunity of recovering their composure, even if the interview had already finished.

Notes were made immediately after each interview including observations about the setting such as who was there, the appearance of the person and general impressions (Kvale 1996). These were helpful later in recreating a mental image of the interview and in evoking the general atmosphere.

Doing the interviews left a lasting impression and led to questioning of the best analytic methodology. The experience of talking to people in their own homes and being guided by their interpretation of the research question could only be properly validated in an analytic approach that recognised agency. Narrative analysis fulfilled these criteria and was vindicated as a choice in the way that analysis of language

reconstructed the strength and purpose of the accounts in a way that physical appearance and vocal ability could not always convey.

Gene discovery interviews

The scientists were familiar to me, although my previous work in the Institute involved little scientific knowledge and this was a source of anxiety before the interviews. The purpose of the interviews was not to clarify scientific facts, but to investigate the process of scientific discovery and its impact. However preparation and familiarisation with the gene discovery was necessary to avoid it becoming "a pleasant social encounter whose content has little or no bearing on the intellectual puzzle which the research is designed to address" (Mason 2002, p. 67).

There were significant themes that arose immediately and entailed reflection for future interviews. These related to the way relationships and networks were a topic of equal importance to the gene discovery. By responding encouragingly when scientists referred to being unused to talking about their work I hoped to gain insight into scientific culture.

Semi-structured interview was chosen to ensure that the interviews were not dictated by routine responses to a familiar situation and allowed me to move back to interesting areas. The aide memoire provided confidence, particularly in the early interviews, where I found the situation threatening in terms of seniority and expertise. Interviewing elites is a recognised genre within qualitative research. The interview with Prof focused on the experience of scientific discovery, the impact of success on career, and the everyday work of practising medicine and science in the rapidly developing speciality of medical genetics. He agreed to see me several times informally over the course of the project where there were particularly interesting or puzzling entries in the medical record. He commented on how he himself had not thought about some of these things for many years and this contributed to the life history and biographical perspective of his contribution. Prof was aware of the possibility of my previous work as a support worker when he was head of the Institute influencing the research but, particularly in the informal meetings, the conversations about details from the past became unselfconscious and moved beyond an awareness of seniority and hierarchy to a relationship with the data.

Spradley (1979, p. 79) describes "a sense of uncertainty, a feeling of *apprehension*" at the beginning of an interview. Making the familiar unfamiliar was challenging, as meeting the professionals was more difficult than if they had been unknown. Reflexivity was required to put the situation in context and awareness of interviewing skills was essential in order to focus, rather than being diverted by nervousness and anxiety.

Listening actively was key to maintaining momentum. The scientists were confident within their field and it required concentration to keep the focus on the less familiar focus of the everyday experience of scientific work, rather than a familiar delivery on the science itself. The exception to this was the lab technician whose interview was grounded in the everyday, local impact of doing the research. This interview was more friendly and chatty, perhaps due to personal similarities of age and stage of life, and here the effort was put into not diverting from the research topic. All but one face-to-face interview took place in work settings and the timing was appropriately monitored. One took place in a home setting and kept to the agreed duration due to travel constraints.

Informed consent was obtained prior to each face-to-face interview. In the case of the online interview there was email confirmation of willingness to participate. The consent forms were sent by post at the same time as the initial interview question and the scientist asked to return two signed copies. There continues to be controversy over the obtaining of signed consent for online research (Johns et al. 2004). In this case the decision was taken that Bill's email indicating willingness to participate was adequate to begin the project, but that signed consent was required before data analysis.

A potential drawback of Internet based research is the lack of commitment and the fact that "Collaboration rests...on human willingness to co-operate over long distances" (O Dochartaigh 2002, p. 110). Bill's interest in the project, combined with previous acquaintance working at the Institute, were both factors in deciding to pursue this research method.

The initial plan of asking questions one at a time, and incorporating issues related to replies into later questions was changed to a schedule of questions due to the time delay in receiving a response. The convenience of asynchronous email is set against the lack of necessity for an immediate response. The sending of the email does not occur in a designated temporal space but instead the email arrives randomly amongst others. The face-to-face interviews were all carried out at pre-arranged times and adequate time was given to them. The lack of exclusivity associated with email may have contributed to the delay in response. The schedule of questions was sent with a note explaining that it outlined areas of interest to be explored by Bill rather than a strict or rigid format. This resulted in a detailed response.

Accessing the medical record

Working with the DM archive entailed negotiating access to the records and physical space to examine them. Previous familiarity with staff and awareness of how busy the department was and how little space was available resulted in easygoing negotiations with an NHS administrator, and I worked during times when desks were available or temporarily unoccupied. Access was given on an ad hoc basis as getting a key would have meant extra administration for the staff. The reality of the DM archive was different to the formality of its title. It has only become referred to as an archive since this project and it continues to function as an everyday part of the clinical service to families with DM. The key to the locked cupboards was kept with the secretary attached to the muscle clinic and she was always helpful about any queries relating to the notes.

Many of the medical records were old and large as they contained information and correspondence dating back over several decades, and concerning multiple members of one family. Physically handling and navigating them was challenging, as the chronology of the information wasn't always apparent. There were also delicate items such as old handwritten letters, and in one case an extensive family tree drawn up in pen and ink by a family member and comprising many sheets taped together. The general format of the records consisted of referral and biographical forms, family tree and handwritten clinical history sheets at the beginning of the notes. Laboratory and clinical result forms were taped at the back and all typed correspondence filed from back to middle reading from oldest date to most recent.

Administrative work had begun to divide some of the largest files into separate files relating to each branch of the family, with each file retaining the original number but suffixed by alphabetical index.

The medical record had to remain on site and could not be photocopied for reasons of confidentiality. A pilot study of five medical records using pen and paper to write down all observations was undertaken. This was time consuming but fulfilled the purpose of gaining as complete a picture as possible of the research focus. The practical consequences were that of needing a space to write and typing up the notes later. Once this was completed a DM archive data collection form was designed (Appendix three) and a laptop computer was used to collect the data.

The medical records, whilst representing a composite of DM clinical research and service, each contained a narrative of their own imposed by the reader as interactions between professionals and family members over time were recorded. The purpose of the record was not to establish a narrative thread but to provide a place where all aspects of DM relating to the family could be contained. In doing so processes over time became visible as the record is mostly kept in chronological order. Following the DM gene isolation family members who had contributed a research sample were asked if they would like this tested. Some of these families had records documenting an uncertain diagnosis over many years. The copies of the typed letters were flimsy and before turning the page it was possible to see from the length of the letter, whether the gene test had resulted in a diagnosis or not. It was difficult to read the records as a detached observer as each entry related another aspect of a person's biography. The act of tracing those points in the trajectory of DM and the trajectory of personal biography indelibly marked them as moments of consequence.

Data analysis

Grounded theory implies an iterative study design "in which the results of the ongoing data analysis inform the subsequent data collection" (Kennedy and Lingard 2006, p. 103). Data analysis was ongoing from the beginning of the project and took place in a framework that recognised context to be central to all enquiries about the relevance of the data. The overall theme of analysis is that of adopting a variety of

approaches, as advocated by Coffey and Atkinson (1996), and of always grounding the analysis, and subsequent theory, in the data.

The DM archive.

Analysis of the DM archive was a challenging task of firstly ordering the data and deciding primary themes from the vast amount of potential data. A pilot study of five sets of DM medical records was carried out, followed by a substantive analysis of thirty- seven further records. Finally a single case analysis was carried out on one DM record of particular significance. The case study emerged out of data collection, reflecting Hartley's (1994, p. 208) observation that a case study is "more likely to adapt to and probe areas of original but also emergent theory". The record selected for the single case analysis was sampled at the end of data collection, when themes were emerging, and recognised for its potential as both a revelatory and an exemplifying case as outlined by Bryman (2004). Whilst making no claims of being generalisable the case study preserves the context of the medical record whilst exploring meaning.

The data was gathered by in-depth investigation of the medical records. Codes and themes were not defined in advance as the purpose of the pilot study, in addition to testing method, was to establish key lines of inquiry that could then be applied to a greater number of records. Analysis was based on interrogating the notes made from the records, rather than the records themselves. Visits to the archive over several months were necessary before a comprehensive collection of the data was completed. During analysis the archive was re-visited to check details and confirm that the coding did relate accurately to the medical records.

The medical records, varying in size from several to several hundred pages, were examined sentence by sentence. Data collected on the DM data collection form developed from the pilot study was inputted directly into Atlas.ti and coded and managed using the software (Lewins and Silver 2007). Codes related to both content and context. Each entry in the records was coded in relation to the personnel involved and the type of activity. This resulted in eighty- three codes for content and context. A separate code was reserved for collection of language relating to emotion

and linguistic devices. Codes were then put into three categories relating to three main themes.

Silverman's (2004) guide to analysis of texts was instructive in formulating the research design. He cites content analysis and analysis of narrative as ways of analysing texts. Coffey and Atkinson (1996) counsel against being constrained by narrow methodological choices and this thesis utilised a combination of methods.

Meaning can be established at a surface and deeper level (MacDonald 2005). Simple content analysis of the number of different professionals who appeared in the records gave an indication of the possible interpretations there may be of the disease, and served as a reminder that all these representations relate back to an individual with their own reality of living with DM. Some of these representations of DM have been given greater prominence and documentary analysis allowed other traces to become visible.

Language and narrative were key areas of inquiry in the study. Particular attention was paid to how the condition of DM was defined and refined over time, as the records included contributions from family members as well as from professionals. Language in relation to social processes was also noted as several of the medical records spanned decades and contained references to changing ways of addressing patients and professionals. Coding for language and emotion was carried out by noting linguistic devices on Atlas.ti but by then manually analysing this large analytic subset. Atlas.ti acted as a data manager in this instance, enabling large sections of data to be referenced for analysis of the narrative context.

The data was also interrogated manually using the analytic strategies of asking what different kinds of activities were going on, what were people trying to achieve, what language was being used, what assumptions were being made and the real effects of what people said as well as what they did (Riley 1990). Motivations and justifications for behaviour were analysed in addition to exploring the nature of the topics recorded. (Riley 1990) The concern was not with true or false accounts but on how the documents give insight into the practice of professionals "while

acknowledging the structural constraints to which they must respond (Silverman 2001, p. 279).

The concept of "networks" and "circuits of communication" (Prior 2004b, p. 386) was central to analysis. Context and networks provided a framework in which to analyse DM as a dynamic concept or "agent" (Prior 2004b, p. 388) in its various representations. Context also applies to the audience for whom the document is intended (MacDonald 2001, p. 199). The dynamic nature of the DM record as "...an entity which is shaped by the context in which it is produced and, in turn, the text shapes that context" (Pettinari 1988, p. xiv) was explored. The approach to the data was interpretive with the aim of investigating " *How* documents are constructed as distinctive kinds of products..." (Atkinson and Coffey 2004, p. 60).

The four criteria of authenticity, credibility, representativeness and meaning as advocated by Scott (1990) formed a useful basis for analysis. Authenticity and credibility in a contemporary document that is subject to guidelines for practice were easy to establish. The process of analysis recognised that here are many people with DM who are not represented in the DM archive, perhaps because they do not subscribe to the model of hereditable illness or because the symptoms in their family are as yet subtle and have not been diagnosed. The remit of this thesis was bound by the archive, and analysis took place in this context but recognising that documents are of interest for what they omit, as well as what they contain.

Single case analysis

Analysis focused on a narrative reading of a key set of notes. Methodologically this was justified because the DM medical records follow a chronological outline and retrospective reading imposed a narrative structure on the way the lives of the family intersected with professionals through the relationship with DM. Theoretical issues associated both with using narrative analysis and with selecting a single case study have been acknowledged. Elliott (2005, p. 98) describes how a "distinction is frequently made by authors with an interest in narrative between 'narrative' explanations as opposed to causal explanations". The use of an analytic framework such as Labov's (1997) places the narrative within a specific context, while Polkinghorne (1988, p. 11) stresses the richness of a narrative account and the way in which "narrative is a scheme by means of which human beings give meaning to their

experiences" Others such as Abbott (cited in Elliott 2005, p. 99) argue, "...it is important not to conflate narrative analysis with single-case analysis". The aim of this analysis was to utilise a rich data source with a lengthy chronology to explore how research was accomplished. According to Murray (2008, p. 120) this interpretive phase highlights "how the elements in the narrative are linked together...what issues are the main themes, what images and metaphors are used, and what are the underlying beliefs and values".

The aim was to use narrative theory "to impose patterns on the past in order to tell stories about it" (Martin 1986, cited in Cortazzi 1993, p. 84). Cortazzi (1993, p. 85) asserts that three main criteria of temporality, causation and human interest must be satisfied in order to define a narrative.

Temporality, or the unfolding of a sequence of events in time, was interpreted as the sequence of events over a career and over the lives of three generations of the same family. Woven through this was the evolving definition of DM in an academic and scientific sense. However the everyday manifestations of the disease and its inevitable deterioration were captured in the details that tracked interaction with the family from the referral of Ben, a toddler with DM, to his death and the death of his mother Alys, and grandmother, Delia.

Causation was implicit in the natural history of the condition. The lives of the family established temporality, and the effects of DM on these lives influenced their progression. Narrative was seen in this study as "not simply a succession of recounted events, but an interesting intelligible whole, where events are connected by time and causation" (Cortazzi 1993, p. 86). Arguably looking at the medical records in a way that gives them identifiable narrative themes fulfils the criteria of "human interest". The medical records are fascinating repositories of detail and are likely to be interesting to a wide range of people. Coding of the records led to themes as diverse as the development of technology, the way in which a disease classification is organised and the changing cast of actors in the records over time. In examining the medical record as a whole it was impossible to ignore the effect that all of these issues have on the lives of the people they document and the analytic aim was to capture that narrative trajectory.

Exploring the medical record utilised reception theory, and the idea of interpretation of the text, rather than looking for a particular truth. The reader as investigator is central to producing meaning and this meaning "is reached through an interactive process that take place between the reader and the text" (Polkinghorne 1988, p. 96). This can allow for a range of possible meanings, points of view or "horizon of expectations" (Popper 1972, pp. 345-347) to emerge. Coffey and Atkinson (1996) have stressed the importance of not making assumptions about texts being representative or illustrative of a context by themselves.

Analysis aimed to allow for the emergence of themes rather than presume that there was a singular truth or story located in the data. The medical records, while they allowed privileged access to a particular context did not define that context. This is particularly true in relation to the families, as they were generally not represented in their own words in the notes.

The analytic focus in this single set of notes was their function as an exemplar of the work documents do through networks and relationships in accomplishing phenomena such as classification. The chronology in this medical record illustrated how these themes were accomplished, and how they changed and impacted on one family over three decades. The impact of gene discovery was reflected through changes in process but the interaction with the family remained grounded in relationships over time.

Gene discovery analysis

The interviews were seen as a process rather than an event, with the initial stages of thematizing and designing following onto interviewing and transcribing. This helped to ensure that the end stages of analysing, verifying and reporting were grounded in the appropriate methods (Kvale 1996).

The use of the voice recorder had practical and theoretical implications. Riessman (1993) described the following five levels of representation in the research process: attending, telling, transcribing, analysing and reading. The stage of transcription involved the transformation of the spoken word to text. According to Mischler

(1991) it is impossible to give a true representation of spoken language and transcribing speech is itself an interpretive act. The aims of the research project guide the decision about the level of detail required, as does the theoretical viewpoint of the researcher (Ochs 1979).

The initial focus of interest in the interviews was the narrative structure: the way in which the scientists told their story and the story they chose to tell. Transcription did not include every pause or emphasise intonation, such as a conversation or discourse analysis would require. The emphasis was on the language used to build and perform the story. Nonlexicals such as "aha" and "mmm" were transcribed in the initial pilot interview but following initial analysis it was decided not to include them in further transcriptions. Although their importance is recognised in relation to interpretation of the text by a reader the meaning making in this project was associated with the way in which the scientists represented their experiences through narrative.

The interviews were transcribed and coded manually shortly after each interview. Analysis of the data took place in the context of grounded theory as a research methodology. There was no assertion of the truth of any account of genetic discovery and the emphasis was on theory generation grounded in the analysis of the data (Glaser and Strauss 1967). Development of theory arose from examination of the data and was inductive rather than deductive.

The principle of constant comparison was fundamental whereby as "incidents or issues of interest are noted in the data, they are compared against other examples for similarities and differences" (Kennedy and Lingard 2006, p. 104). Although there was a general interview schedule referring to broad areas of the topic, initial interviews and their analysis did inform subsequent interviews.

The major stages of analysis were carried out by concentrating solely on the data. The initial transcript was coded for verbs and adverbs to give a sense of the underlying narrative structure. The data was then coded and cross- indexed as themes began to emerge. Following this the data was analysed closely for narrative features such as metaphors and similes. This was an ongoing process during which emergent themes were checked for exceptions as well as for similarities. Another aspect of

analysis involved examining closely how the events surrounding the time of gene discovery were described and the data was also interrogated to look at overall narrative plot and narrative trajectories from the initial description of gene discovery. The interview transcripts were also coded and managed using Atlas.ti, which was particularly useful for referencing data for quotation.

The analysis could be described as occurring in layers rather than along a continuum as theoretical concepts emerged from the data. The first application of theory was related to initial analysis that had been done intuitively as a way of exploring the data holistically rather than breaking it down. Riessman (1993) recommends this as a first step to becoming familiar with the data. The initial analysis resulted in a basic storyline analysis to which the structural concepts of Labov (1997) applied. This allowed the underlying narrative structure to become more obvious but was not considered to be a definitive analysis.

Analysis of narrative function followed, looking at what purpose the narratives were fulfilling in terms of their underlying messages. Moral tales, career chronicle and tales of success and motivation were explored using Burke's theory of dramatism (1945). Ginsburg's (1989) adaptation of Sklovskij's framework of temporal sequence was then applied resulting in an in-depth focus on the narrative plot. This was based on the analytic observation that, although gene discovery was the agreed topic of exploration, the narrative trajectories reflected its significance in relation to many other perspectives such as career and legacy, rather than simply a description of the events as they occurred. This analysis allowed differences in narratives to be explored as well as similarities.

Analysis of metaphors and other tropes was used to look at how scientists tell stories and how they construct narratives. This explored descriptions of the scientific world and how the unfamiliar is communicated through culturally available forms of language (Coffey and Atkinson 1996).

At this point in analysis there were many emergent themes that were strongly grounded in the data and reflected recurring focus in the narratives. Themes such as competition and collaboration, legacy, memories and scientific identity offered

insight into the scientific world and in particular the world of clinical genetic research. Accounting devices such as repertoires of luck and judgement in scientific discourse, contrastive rhetoric and accounts of justification were then explored (Mills 1959; Gilbert and Mulkay 1984). This highlighted the emotional content of the language used, resulting in a further analysis focused on the context of this emotional register and its significance in relation to the personal meaning of the impact of gene discovery for the scientists.

Family interviews

Data analysis should not be left as a consideration for the end of the process but should be a constant reminder of the underlying purpose of the interview data collection. Seale (2004) suggests asking whether the interviews are an exercise in data collection or data generation. His definition of interviews as being a resource or report of experience, or as an event in its own right was a useful starting point.

Data management was started before collection with careful consideration of possible questions or significant issues. The organisation and ordering of the data in a consistent way was essential for data retrieval and a continual "moving back and forth between your intellectual puzzle, your research questions and your data" (Mason (2002, p. 159) is recommended. The status of the data is one which attaches multiple meanings and which may be read literally, interpretively and reflexively (Seale 2004). Literal readings may be significant in talking about shared events such as the clinic as seen from the point of view of the clinician and the patient. There was not a true or false dimension to the accounts but rather an appreciation of accounts as "displays of perspectives and moral forms" (Whyte 1980; Gilbert and Mulkay 1984; Silverman 1993, p. 107).

Themes emerged as the data was analysed, which informed further interpretive analysis (Glaser and Strauss 1967; Coffey and Atkinson 1996; Charmaz 2002). The data was analysed using both manual and Atlas.ti software techniques. The analysis was based in grounded theory, where each analytic step informed the next. The semi-structured interviews were organised using cross sectional and categorical indexing. The interview analysis followed the preliminary analyses of the medical records and the scientist interviews so there were already some indexing categories apparent but

it was important to stay open and flexible to new and emerging themes (Mason 2002). A data display method is a useful way of keeping a template of themes and this utilised Atlas.ti at a basic level (Miles and Huberman 1994, cited in Coffey and Atkinson 1996).

The analytic strategies described in the scientists' interviews were applied to look at narrative structures and accounts. The grounding of all the family interviews in everyday experience and resistance to a disease model of DM was explored. The scientific interviews were then reassessed in the light of emergent themes from the family interviews. Narrative interpretations of what constitutes DM, how scientific progress was experienced and perspectives on DM were all compared.

The approach to the interviews was one of active meaning making as a collaborative process (Garfinkel 1967; Sacks et al. 1974.). The aim is to show "both the *hows* and the *whats* of the narrative of lived experience" (Gubrium and Holstein 2002, p. 78).

Summary

Lack of motivation and apathy has been associated with DM (Olsson 2002). While this thesis fully acknowledges the influence of physical weakness on the length of the interviews and quality of the recordings, acknowledgement is due to the families who gave generously of their time and energy.

Recruiting from established registers has associated considerations such as the possible increased motivation of people who agree to be included on one. There are many more people with DM than those who attend muscle clinic so there is a group of people who are not represented in this research. However of those people who agreed to take part there was variation in age, extent of disability, family background, nature of referral to the Institute and social situation in terms of employment and marital status.

Research design was governed primarily by the aim of maximising the potential of the rich and varied data sources to extend already documented knowledge about DM and gene discovery, and to explore how this scientific success translated into everyday practice and experience. The aim was to explore the impact of scientific

progress through the experiences of the key actors. It was hoped that in this way the significant issues would arise as they were relevant for the scientists and family members rather than dictated by me. Constant review of the methods resulted in perspectives characterised by depth of emotion in both the scientific and family interviews and reinforced by the different approaches taken to data collection and analysis of the DM archive.

The methods were more than the vehicle for carrying out data collection and analysis. The data itself influenced analytic choices. There was an active and iterative process of moving between the data, emergent themes and consideration of method. Use of grounded theory meant that there was real learning from the data, rather than the application of fixed ideas to it. This subtle distinction applied to all aspects of the thesis where there was an attempt to be responsive rather than prescriptive.

Chapter Four

Molecular Detectives

Introduction

Well, it was quite tense, in one sense. Certainly when we were getting towards the closing stages.

(Abstract)

...I was working ridiculous hours particularly – sixteen hours a day – and I would often phone up [Tom] and say, before I went home, because he was in Boston which was five or six hours behind and I'd say right I've found X, Y, Z, you know...what have you got...and so he'd do the next bit in that kind of way. Particularly at the very end...we were just like a conveyor belt, just getting the data. And that was a bit mindless in a way.

(Orientation)

I can remember – there was one odd bit and we had this feeling there might be a triplet repeat involved and ...

(Complication)

on the morning that he [Andrew] went up to London I had developed the autorad which showed that...we'd actually got something and they [another group in the collaboration] had found something as well,

(Evaluation)

...I can remember showing ... just to make sure I didn't imagine it and my hands were shaking when I was showing him [Bill] and it was just...

(Result)

After all that work and all that effort, to realise you've actually got something.

(Coda)

Pauline, post-doc scientist on local gene isolation team 2008

Six scientists reflected on the impact of the 1992 DM gene discovery during interviews carried out sixteen years later. Their accounts of discovery narrated the relationships, emotions and personal meaning of being part of a small local team in addition to the dynamics of interacting with the international collaboration within which they played a distinct and key role. The way this was illustrated, through anecdote and use of metaphor, gives insight into the world of the research scientist. Through their narratives and linguistic choices they reveal a highly social and complex world made accessible through colourful language and challenging the stereotype of the isolated scientist and the "eureka" moment. Moments of discovery must be validated by others within the scientific community and are further analysed beyond the laboratory and in the clinic in terms of what they have to offer

to the clinician and the family. Descriptions of "figuring it out", "false leads", "red herrings" and "letting the cat out of the bag" evoke mystery, suspense and detective work. A unique phase of scientific research, where being first to discover a gene was the only achievement that had relevance for those involved, is illuminated.

The team told narratives of motivation, turning points, boundaries, relationships, gender, personality and the personal meaning of gene discovery. In doing so they spoke of how a group of individuals were shaped into a team through exploration of a hypothesis. This hypothesis was shaped by technology, luck, hard work, and the social processes of competition and collaboration. Emotions of ambivalence and regret were narrated alongside pride in contributing to the scientific canon, and the importance of relationships within the scientific world was reconstructed through accounts of the personal meaning of the discovery.

The pseudonyms are Prof, the team leader and clinical geneticist who had overall leadership of the Institute, Andrew, a senior scientist, two post-doctoral fellows, Pauline and Tom, a laboratory technician Kay, and Bill, a PhD student, who gave his interview by email.

Laboratory Cultures

Creative Accounting

The scientists gave accounts constructing the field of clinical genetics as innovative. This was accomplished through descriptions of an "exciting project" with new scientific techniques. In doing so they portrayed a culture that was distinct and dependent on personal characteristics as well as a particular focus of work. They contrasted the research scientist in genetics with other types of scientist and used metaphors highlighting creativity, flexibility and resourcefulness. Processing and remembering large amounts of data was part of other areas of science "whereas with genetics you had to think." The scientist brought these characteristics to the DM laboratory environment, which was recognised as being a progressive setting.

> *I built equipment which took about a day and then spent a year learning to play with DNA. I loved it as at that stage it was a new field and as a more classically trained biologist colleague put it my experiments were of the F**Kwit physicist type that fortunately for me worked. I got a few papers out of this work.*
>
> *Bill*

The scientists constructed accounts that drew on a vocabulary of innovation and practicality. They narrated the steps by which hypothesis and scientific ideas are made visible and communicated to others by means of making physical representations or models. The scientists reconstructed the bodily work that was involved in these representations and enacted the role of the scientist as engaged creatively, physically and emotionally.

Becoming involved with genetics constituted a statement about an approach to science and personal strengths. The accounts stressed chance and following up a potentially exciting possibility as a way into the DM team.

> *I went to a lecture...this was in the early days of human genetics, the DNA sort of stuff and he was very enthusiastic and I was looking for something to do so I thought I'd go and have a chat with him after the lecture and he gave me the phone number for...He said there was this bloke [Prof] who was looking for some new staff and give him a ring. So I did and I got an invitation to an interview.*
>
> *Andrew*

> *I was looking for the next stage and I saw this job advertised...which was more on the human side of things...well a lot of these things happen by chance...it was a very exciting project involving a lot of exciting genetics and also modern techniques that I hadn't done before.*
>
> *Pauline*

Creativity and inventiveness, along with a hands-on approach, distinguished the DM research scientist. Recurring descriptions of a relaxed atmosphere where there was openness to ideas constructed an intellectual space in which originality and possibilities were encouraged. The way this work was carried out was illustrated through metaphor and contrastive rhetoric.

> *We were working it out. We were figuring it out. We spent a lot of time thinking is this approach going to work and doing it and it either did or it didn't. So we developed technology. I used to have a soldering iron and I used to make things and I used to wire them up to computers to make them run properly. Stuff like that you just wouldn't do these days. Because it probably isn't safe for one thing...It was really bread and butter stuff that you sort of improvised and it was more a sort of cottage industry thing.*
>
> *Andrew*

The importance of modelling and constructing hypotheses was conveyed through language such as "figuring it out" and exemplified through reconstructions of experiments. The use of equipment that "probably isn't safe" served the dual function of contrasting the pioneering and inventive spirit of the past with the regulated present, and the privileging of imaginative use of limited resources over

advanced technology. Two of the team specifically mentioned the current format of the university Research Academic Exercise as a reason for constraining the type of creative thinking that had led to discovery in the past. Andrew contrasted this with the present where "...we're all meant to be pretty much the same sort of person. They've got the business model of how they want people to be." The past approach in the wider scientific world of allowing for some speculative research in addition to more standard work was reconstructed in the everyday perspective of the DM project. Narratives privileging the past placed the gene discovery in the context of a unique era.

The importance of contact with families as a source of samples was narrated across all the accounts. The "good relationships between the clinicians and the molecular researchers" was recognised as "a major strength" of the team. Perspectives on the families with DM varied amongst the scientists and reflected back to the nature of scientific work.

> *I think you're in science because you're not a people based person. I think in all honesty...I think you know where your boundaries are ...Because some people will always do both, will cross the boundaries...But it's interesting how many clinicians start off doing medicine and then end up doing a non-patient based job...*
>
> *Kay*

According to Prof the scientists "were very interested [in the clinical work]. But they didn't feel that was their work. They were quite happy to be kept in touch and grateful that there was a good source of, you know, material." Prof and the scientists shared curiosity about the biological phenomenon of DM. This was noted with ambivalence in a family interview where Maria, who has DM, commented that, "it [DM diagnosis] is such a bombshell and that, I've always thought, was a bit cruel, because to them [the researchers] it was so interesting".

In interviews carried out with members of the DM team in 1992 Batchelor et al. (1996, p. 235) observed:

> The stakes are high in human genetics – it is perceived by many in the scientific community we studied to be an area of high status, and there is substantial prestige in being the author of the first publication that describes the cloning of a particular disease gene.

The contemporary DM gene discovery narratives highlighted the importance of succeeding in a venture where the outcome could determine the next stage of a

career. This was common to all the accounts, even where there were differences in the personal meaning of the gene discovery. This related to the specific nature of the research, which was described by Prof as "an unusual phase...it was collaborative and it was very, very dependent on collaborations but it was unlike most of the work before and most of the work after." Collaborations of groups, like that including the local DM research team, competed against each other to identify the genes for single gene disorders. The way this was accomplished involved intellectual and technical skills but also negotiation of resources and personalities. The possibility, narrated by Pauline that "somebody could have spent years...you know, working on something, and suddenly I could pick up that last little bit..." acknowledged the role of opportunity, politics and luck. The predatory imagery conveyed the risk involved and the speculative nature of aspects of the endeavour beyond the immediate bench-work in the laboratory.

The end result was not gene identification alone and resulted in recognition of the scientific advance and the successful personnel in the published accounts. Prof recognised that publication was important to the "people who are either doing a PhD thesis whose thesis depends on it [gene discovery], ...people [scientists] who are doing a post doctoral fellowship whose next post depends on it and people on the clinical side where you know their career is influenced by it."

Kay gave a different account. Whilst recognising the general importance of gene discovery for a scientific career this was contrasted with the nature of her own work. Internal recognition for satisfactory accomplishment of the everyday tasks that kept the laboratory functioning was more relevant than external recognition through official scientific publications.

The accounts of the impact of gene discovery on career thus functioned at both the professional level of career development and the everyday level of personal work satisfaction. They opened up the hierarchy and roles within the team allowing different perspectives on recognised interpretive registers such as competition and collaboration and hard luck and judgement to emerge (Gilbert and Mulkay 1984; Atkinson et al. 1998). The unique phase of research was contrasted with how "the nature of the work's changed over the past ten or fifteen years", and investigating

the complexity of genetic changes has replaced the phase where "...in DM you were looking at these really quite gross changes really." The career of the research scientist did not relate to professional stages or grades but to the potential of a project to lead to career development. All of the scientists narrated their work in terms of personal meaning although this varied according to their position within the team. Kay narrated a career that was also bound by the routines of the laboratory and was subject to disruption in major events such as gene discovery.

Being Grounded

Curiosity and an interest in solving or finding out more about a phenomenon justified a work ethic that did not focus on theory or reflect back on past achievement. The research scientist was constructed as someone who was involved "on the ground" and who engaged actively with problems rather than with theories. Reflection was contrasted with doing. Accounts of being a scientist constructed a combination of creativity and practicality. The importance of looking forward was justified as part of the inexorable progress of scientific knowledge, whereas the personal activity of reflection was a cause of discomfort.

> *...it was and continues to be a career that I have not evaluated in much detail so answering your questions has made me a bit uncomfortable.*
>
> *Bill*

> *I haven't heard many scientists talk about that kind of thing anyway...they don't...They're much more interested in what they're going to do next...That's part of the mindset*
>
> *Andrew*

Reflection as a discursive practice within scientific culture was not prioritised and did not constitute part of the scientific "mindset". Prof also spoke of being aware of "paradigms" and theories about how science is accomplished, but of being a person who had "just been involved with it and done it". The past was important in the clues it might provide for the discovery. In this way the phenomenon of anticipation was "chewed over" as there was "the feeling it's trying to tell you something". Reaching understanding and contributing knowledge that might lead to further scientific discovery or clinical application provided motivation, as did the excitement of discovery but "there's not much theory for people on the ground". The DM team made a significant contribution to the success of the collaboration through the knowledge of DM relationships gained through close clinical contact with families.

This reference to being "on the ground" and working from in depth knowledge of the condition was stressed by other members of the team. Being a good scientist was not effective, according to Tom, "if you didn't have someone who was the critical link between the patient and the researcher."

However there were key differences between the technician, the clinical geneticist and the scientists. Being a scientist did not mean being part of a homogenous group, a point which the scientists illustrated through constructing defining characteristics of the type of person likely to research genetics. The career of the research scientist was dependent on the success of projects.

> *There's a lot of debate goes on, even now over who's where on papers because at the end of the day it does make a huge difference to somebody in where their career is going...*
>
> *Kay*

The career of the technician was dependent on the smooth everyday functioning of the laboratory, while the clinical research career involved crossing boundaries between service and research. The first reference to other careers in science came from Kay discussing her relationships with other laboratory staff outside the DM team and the benefit of having a mixture of people "so you weren't all stuffed into the same place with tensions between things or anything like that". The specific attributes of the research scientist were again contrasted against other possible career options.

> *...science doesn't only work in research. You can go into industry...I mean research isn't the only formal scientific career. Research works for a lot of different people. They like the environment. It gives them the hours, doesn't give them the pay, depends on why you're doing it. If it's an innate curiosity, which people like [one of the DM scientists] have, then yes it's nice to have all the papers but on the other hand that's not, I'm sure that's not what's driving him. He's got to be focused on all the other things. On the cow with six udders or whatever it happens to be...I think it's a star [finding the gene]. It's the golden star in a career.*
>
> *Kay*

The example of the extraordinary as a motivating force, alongside innate curiosity, again reinforced the research scientist as an unusual and lateral thinker. The possibility of the great discovery is privileged above the recognition in publications, serving to highlight the inner rather than external motivation of the researcher. The

imagery of the golden star highlighted the significance of the achievement within a scientific research career.

Prof was the only male member of the team to draw on the personal domain as a factor in achieving success and enabling the coordination of activities and input that extended beyond the conventional working day.

> *More of a problem was...how do you fit all these different things? And the answer is they didn't really fit in and I suppose that's where having a fairly tolerant family is necessary. Because these things don't fit, do they? To the conventional working day.*

The importance once again of being embedded in reality is communicated through the use of "down to earth" reinforcing the moral lesson that personal character and ability to remain grounded in reality were more important than becoming waylaid by hierarchy or success.

Chemistry lessons

Prof stressed his role as someone who did not get involved in the day-to-day running of the laboratory. He explained how he had learned the necessary techniques to be aware of the laboratory work but that he did not carry out bench work. Following the scientific lead rather than a set management protocol privileged the central importance of the research question and the allocation of resources towards its solution.

> *I spent time at [another lab] and I'd go up for a couple of days each week for quite a long time but to be frank the value of that was not that I learned them particularly well but it helped me to understand what was going on...*

In contrast Kay's role as technician involved negotiating dynamics within the laboratory and accomplishing the work necessary to keep the day-to-day routine functioning smoothly.

> *They were quite focused because everybody was working on the myotonic and working towards it...Everybody was sort of working towards that goal. I probably less than the others. I knew which diseases I was working with and I was happy with the technical side which is what I enjoy doing. But I probably wasn't as focused about it as obviously perhaps, you know, [Tom, Pauline and Andrew], because that was kind of obviously their baby really. Whereas I didn't feel quite as much attachment to it I suppose.*

Gene discovery was potentially a disruptive element in the laboratory, as was the growing tension. This tension related to personalities and to focus. The nature of

this phase of clinical genetic research meant that only the team to identify the DM gene first would gain recognition. Being part of an international collaboration increased the possibility of success for a small local team such as the DM team at the Institute, but within collaborations there were also tensions relating to sharing of knowledge and resources. Within the DM team itself there was tension between the scientists as the pressure mounted, and also pressure on Kay, whose motivation and focus related to maintenance of the immediate environment of the laboratory. The detachment of the language reflected Kay's point of view "As a mere minion at the bottom". Kay and Prof both referred to changing dynamics but their perspectives reflected their differing contributions to the team. Kay's use of domestic imagery strengthens the communication of the routine but essential tasks that made up her work. Internal dynamics in the laboratory related to "juggling personalities" and political struggles within the team relating to feelings of inclusion or exclusion. The exclusive focus on gene isolation was contrasted with the ideal state where "you should be thinking ahead already" to the next phase of work. Prof also referred to politics and dynamics but in the wider context of collaboration and competition. Control for Kay was related to the ability to carry out the laboratory work well, despite personalities. This contrasted with control for Prof, which was located beyond the laboratory and involved active engagement with personalities "to try and persuade people that by sharing and collaborating as fully as possible everybody would gain rather than just one person." Although the DM gene discovery was published in 1992 several of the accounts referred to the possibility that tension relating to the exact timing and contributions of teams to the collaboration still exists.

Kay and Bill referred to hierarchy within the team in relation to how their status protected them from politics and ensured that "You might get a fallout...but you weren't actually in the cross fire between any of the politics." The sense that hierarchy mattered less and politics more as the scientists became more senior was reinforced by Kay's observation that in the DM group sixteen years before "there was more of a sense of hierarchy than I have now but maybe I'm in a different place in the hierarchy [now] so I don't realise what's going on." An anecdote of "the mighty boss" [Andrew] making a mistake in showing Kay how to set up an experiment conveyed the moral that "if you're calm and collected about everything

then everything is generally fine. It's when you get stressed about it that things go wrong." The importance of mastering techniques and maintaining a calm atmosphere was contrasted with the challenges of managing dynamics within the team.

These dynamics had a common theme across the scientific accounts. Emphasis was placed on intangible aspects of how the group worked as a team, rather than descriptions of management. These accounting devices were described by Gilbert and Mulkay (1984), who contrasted the empiricist repertoire of scientific discourse used to portray the public representation of science as factual and logical, with the contingent repertoire used privately by scientists to describe the less predictable processes of everyday work. The DM scientists' accounts extended the contingent repertoire by using it to reconstruct processes reliant on human actors in an emotionally engaged way. Prof's overall management was analysed in terms of good strategic decisions relating to scientific progress rather than personnel, reflecting the reluctance to narrate less tangible aspects of how progress was made but accomplishing this through positive attribution rather than blame.

> *...is a good leader in genetics. He set clear boundaries between clinical and research work (in my experience) and recognized that they function in different ways.*
>
> *Bill*

> *He was very good at seeing what was going to happen next and getting organised to deal with it.*
>
> *Andrew*

As with all of the team dynamics hierarchy was narrated as flexible, depending not only on roles within the team, but also on intellectual capital. Andrew, the senior scientist, referred to the need to know what junior staff were doing and how "they wouldn't have set something up on their own" or they would have got a "a slapped wrist...Unless it was a really brilliant idea of course." The importance of allowing the opportunity for serendipity was narrated as an important aspect of the team. Bill described having to prove his skill to Andrew by using the rhetoric of sport and challenge to describe how "we had an arm wrestle but in the end he was happy to leave me to it".

The language used to describe the day- to- day interaction within the team focused on the use of personal resources to negotiate relationships rather than actively

developing cohesion. These relationships developed independently of management strategy and were related to inner qualities that facilitate good relationships and make it possible to deal with more difficult situations. This use of the contingency repertoire (Gilbert and Mulkay 1984) functioned to explain difficult interpersonal situations and to associate management of scientists and teamwork with personality or "chemistry" rather than with formal management strategies. This served to protect the flexible environment in which intuition and creativity were not hindered by rigid structures. In doing so it also functioned to privilege personal responsibility and diminish accountability where a lack of definition did not formalise rights or responsibilities. This reinforced the earlier observation that the contingent repertoire was not used solely to counterbalance the empiricist repertoire of objective infallibility. The DM scientists used it to illustrate how these processes, governed by human actors, functioned. There was acknowledgement of positive as well as negative explanations for behaviour but a characteristic reluctance to analyse dynamics within a management framework.

> *A lot of it is personality. It's interesting. It's to do with personality...And [DM scientist] wasn't confident enough to ride it out and say 'Tough. I don't care'. So there were things like that going on and just juggling personalities because I mean [DM scientist] wasn't the easiest person to get along with but I mean I seemed to get on all right. As long as you knew what people are like.*
>
> *Kay*

> *I think it's [management] pretty chaotic actually. I think it works from the bottom up. I think its chemistry between individuals and what they do. How they behave towards each other and that sort of thing. I don't think there's a whole lot of management goes into it.*
>
> *Andrew*

The code of conduct by which the research scientist operated was implicit and dependent on self- regulation of behaviour. The significance of personality again highlighted the importance of individual characteristics and their contribution to managing the research problem rather than the research team. Narrative analysis of the accounts indicated that, in addition to the empiricist and contingent repertoires as forms of discourse presenting the infallible public face of objective science and the fallible explanation for human error, there was an additional register of emotionally engaged and positive language offering insight into the everyday work of the laboratory.

Gender

Both Kay and Pauline constructed accounts of women in science that highlighted unusual qualities. Being a woman in science was discussed in a way that being a man was not and being a successful female scientist was a challenge that atypical women, or those prepared to make sacrifices, achieved. These accounts drew on aspects of personal life and referenced the world outside the laboratory in terms of domestic and personal relationships. Pauline described how "...initially it was fun...worked really hard, socialised together so it was quite a nice atmosphere to be in...it didn't feel like going in to work."

Whereas mentoring for Bill, as a junior male scientist, related to age and supporting younger members of staff, gender was the reference point for Pauline. The influence of an "eccentric" woman tutor who had became part of academic science via "a very odd route" and was supportive of women in science played a key part in Pauline gaining the confidence to pursue a higher degree and a career in scientific research. The ambivalence towards how research is achieved was a recurrent theme that moved between questions of whether gender or personality was the definitive attribute necessary for success.

Kay narrated success as a combination of gender, personality, and lifestyle choices.

> *I think it comes down to personality and it depends on your priorities. It depends on how focused you are I think. If that's what you want to do then you're prepared to drive that path. What becomes complicated is if you want to stop and have a family...Because at the end of the day you're the one who has to stop, step out six months, nine months. If you're not bothered about having children and you're very focused then I think you can progress very well...The women I've come across at that level are quite interesting characters and they seem to be either extremely stroppy or they have certain personalities...there's a lady in...I know who's pretty successful in her field. She's not ferocious but she's quite scary in all honesty which men don't seem to be. Whether it's because they overcompensate for competing demands or you just have to be that sort of personality. I don't know. I think it's largely personality.*

The drawing on personal resources and the significance of personality is at variance with the objective, detached and impersonal language of public scientific discourse. Science and the scientific career were described in the language of constant motion and as a trajectory that the scientist must keep pace with. Contrasting female with

male scientists highlighted the additional factors for a woman in the field and constructed unusual characters functioning in a challenging environment. Although these explanations served as accounting devices to justify behaviour, paradoxically they did not make everyday scientific work easier for other women scientists.

Solutions through Problems

Problems and the approach to them were a key aspect of all the accounts. Differing perspectives on the nature of problem solving related to roles within the team. Prof and the scientists narrated problems as being an important way of making progress. They were intellectual material to be actively engaged with. The focus on problems for the scientists was specifically related to the scientific hypothesis and functioned as a form of discourse to give insight into how the scientist accomplished work. Problems also served to illustrate the trajectory of the scientists' work, where they moved from one problem to the next rather than reflecting on what they had just accomplished.

> *I'm more interested in immediate science problems that can be solved and then I can move on to the next one. I don't always care what the content of the problem is but I like dealing with it. And solving it. And genetics is quite good because everything is quite problem based, you know.*
>
> *Andrew*

For Kay problems were related to dynamics or management issues within the laboratory, while Prof moved beyond the laboratory into the clinic and the family home to give a wider perspective on the relationship of problems to everyday life.

> *And I remember turning things upside down and saying, 'Right, we can't avoid these problems, there are going to be all kinds of them. Let's make them the main study. Then we can analyse and find out what's the best way to avoid them or get around them. And we're going to hit them whatever happens.' And that did indeed happen.*

Prof's broader perspective encompassed the implication of scientific advance on areas like insurance, ownership of genetic information, and childhood testing. The moral lesson of being grounded in practical experience was again expanded upon to illustrate that the value of contemporary engagement with difficult ethical issues was not theoretical but lay in their importance to families. Verbs such as "hit", "get round" and avoid" constructed progress as a trajectory or journey with problems as potential obstacles. Having the right perspective could turn these potential obstructions into opportunities. The purpose of narrating "a raft of issues" that were

"well and truly anchored in day to day practice" lay in the possibility of being able "to help and not harm the people where it was arising." This perspective moved the excitement of scientific discovery to a moral landscape guided by the impact of new knowledge on families with genetic conditions.

Being part of the DM research team was narrated as a complex balance of personal attributes and ability to keep pace with changes in scientific knowledge and in dynamics amongst individuals. The narrative accounts opened up differing perspectives according to hierarchy and gender but similarities in the construction of the type of scientist who was likely to work in the newly emergent field of clinical genetic research.

Turning Points

The account of DM gene discovery given by Prof was constructed around key turning points. These narrative devices highlighted the theme of the importance of clinical research being grounded in clinical knowledge and experience of DM through work with families. This moral tale occurred throughout the interview and served to connect the turning points narrated at various stages. The foundation for two key turning points of securing funding and access to technology could be located in the initial work of seeking out families with DM in order to build on knowledge about the natural history of DM.

Patronage

The building up of an institution from its beginnings with one geneticist was dependent on selecting the right projects and developing an institutional identity. The importance of a motivated individual taking an interest in a particular condition, rather than stepping into an established framework also marked the field of clinical genetics as new and different to other medical specialities. Prof brought the experiences of having a mentor and learning through apprenticeship to his new department of medical genetics. His recollections of the origins of the DM research gave insight into the way the department would develop.

> *That goes back a long way...The way that came about was that when I went over to do a fellowship with Victor Mc Kusick, not having any clear idea what I might do, his first suggestion was something quite different which was*

to try and look at recessively inherited families with osteogenesis imperfecta. And for a month or so I beavered away through hundreds and hundreds of files...So I went to Victor and said "Look, this doesn't look very promising". And he agreed. And then he said, "Well, look, we've got this project on Myotonic dystrophy just starting".

Returning to the UK to set up a department entailed finding a research interest that was personally motivating but that also fitted into the wider research community. Building up clinical expertise enabled opportunities to be taken as scientific techniques developed.

The classification of DM itself was based on clinical experience. The foundations for this lay in the early work with Victor Mc Kusick where, through visiting and assessing families with DM with a scientist in rural America, Prof began the practice of grounding DM research in clinical expertise. The boundaries between having the condition and not were challenging due to the variation in manifestation and the poorly understood trajectory of DM.

Well that was where the problem started because – well, one of the things which came out of the study was – I think it was 17% of people who were symptomless turned out to be affected. So in other words you would be misclassifying people the whole time as unaffected when in fact they really had mild features. So that was where the skills were needed.

Initially, before the resources to begin the laboratory work were secured, this clinical work was carried out throughout the UK by Prof, and formed the basis of the clinical expertise central to the DM gene discovery. Again the narrative was grounded in personal experience, constructing a picture of the young research geneticist, and using personal biography and the narration of specific skills to convey a unique time and a model of practice.

I wrote round to all the paediatricians in Britain basically asking 'Have you ever seen a patient with this?' And then ended up again going round the whole country, as far north as Aberdeen, visiting these families at home. And I ended up with a series of – I think it was seventy something congenital patients. Which really again provided a big database. Because quite apart from the children of course there was all the families. And so then I wrote that up, two papers in Archives of Disease in Childhood on congenital or childhood Myotonic dystrophy in Britain.

The construction of the DM work, and its shaping into a project resulted from the motivation to take a research interest and raise awareness of the condition firstly within the medical profession and then directly with families. Prof, in his

description of the geography of research, captures this unusual phase of interaction with families and their transformation into the "series" and finally the publications.

Funding

The rising profile of DM resulted in recognition of Prof and his growing department as expert in the condition with direct impact on obtaining resources. The contingent nature of funding was central to the beginning of the DM project itself and Prof conveyed the importance of this in a dramatic narrative.

> *So suddenly I realised, heavens, this gene is on chromosome 19, which wasn't the one that we put in our application.*
> *And so I remember coming back home, rushing in and saying to the girls in the office "Has that application gone off"*
> *And they said "Oh, we're terribly sorry, we've been awfully busy and people being off sick, it hasn't actually gone off".*
> *So I heaved a big sigh of relief. And I remember we got the paper out and we went over it in Tippex and we Tippexed out whatever chromosome we'd originally put in and let it dry and typed in 19 over the top.*
> *And that was then received much stronger and then they funded it.*
> *And I gather that in the end they funded two projects. They funded one safe and one speculative one.*
> *Ours was the speculative one.*

The evaluative function of narrative (Labov 1997) of placing events in a meaningful context highlights the drama and contingency of scientific discovery but also of funding. The drama is conveyed through remembered speech, heightening the urgency of the story. Details such as typing over the drying corrector fluid evoke the past and further add authenticity to the account. The funding of a safe and a speculative project mirrors the scientists' accounts of the importance of the unusual idea in addition to the more ordinary but equally important projects. The resulting funding enabled Prof to set up a laboratory team to provide a scientific basis for exploring the clinical observations he had been building up alone until that point. The ability of narrative accounts to go beyond the taken-for-granted knowledge about a scientific endeavour and convey how that all began grant temporary access to an unfamiliar world. Prof's evocative accounts of the past, in particular home visits to families with DM, place DM gene discovery in a historical context where the past is implicitly and explicitly contrasted with the present. Stories of how the scientists used to build models using equipment that would be considered too dangerous now convey both cultural and personal characteristics, while details such as Tippex locate the narrative temporally in the past. The way "Actors continually reinterpret given

actions as their biography unfolds and as changing circumstances lead them to fit these actions into new social configurations" (Gilbert and Mulkay 1984, p. 9) placed DM gene discovery within the framework of a unique phase of research. Through their retrospective narrative reconstructions of the way activities were carried out, and details of naming equipment and objects no longer used, the DM scientists evoked a way of being a researcher and doing genetic research that belonged to another era.

Technology

In the early 1980's developments in recombinant DNA technology marked a defining moment in the field of clinical genetic research.

> *I suppose it was around 1980 or so when the DNA polymorphisms came in. And this was when a huge change started which we were right at the beginning of. Because...in London had started working on mapping genes with DNA markers...And then we put things together with us providing the families and the linkage analysis. And then initially the DNA analysis was done the London and then moved down to [Institute]...it was at that point that...was very keen to look at something on a different chromosome...And then I thought, well, why not Myotonic dystrophy?*

Andrew, the first scientist to join the new team, spoke of how "the group got bigger each year and then other diseases got added to the portfolio", resulting eventually in the building of a new Institute to house laboratory and clinical services. The way this research portfolio was influenced by the work of other research groups indicated the boundaries that lay behind careful choice of projects. Prof recalled that, "Well, Myotonic dystrophy was never really, like I said, there were never huge numbers of people interested in it."

Through their narratives the team also highlighted the importance of patronage and of leadership by a person strongly motivated to work on a particular condition. This gives a different picture to how research develops than the account of DM gene discovery published in scientific journals. The gene discovery as narrated by key actors was not a straight trajectory but was contingent on testing and discarding hypotheses, choosing collaborations and also on negotiating an identity in the wider research field as experts on DM. Funding and technology influenced the direction of the project. The incremental nature of the research and "actually isolating the

gene was a long way off – people weren't even thinking about that, we were mapping it" illustrates the iterative nature of progress.

Social Scientists

The Conference

The laboratory, the clinic and the home were all places where the day- to- day work of researching DM took place. Narratives reconstructed a cast of characters who served to illustrate how scientific work is carried out in reality, through contrasting accounts of good and bad behaviour and the juxtaposition of luck and hard work. The scientists used contrastive rhetoric to portray the acceptable and unacceptable aspects of scientific behaviour. This was encapsulated in stories peopled by "cowboys", and unprofessional behaviour such as "letting the cat out of the bag" and leading others "up the garden path". Through these familiar tropes aspects of scientific culture were communicated and made visible to a non-specialist audience. This social landscape had cultural values made visible through the attribution of characteristics. The social setting of the scientific meeting or conference represented the intersection between this daily activity and the wider scientific community. Conferences and meetings attended by the scientists were the subject of repeated stories. The conference was not constructed by the scientists as an arena for the development of a professional identity but related to a scientific idea or principle.

> *Most of the work happened in the bar especially in relation to checking out ideas and working out collaborative work. We also built up good friendships through that time which are still strong.*
>
> *Bill*

The conference, revolving around a timetable of talks and presentations, is a recognised academic forum. The scientists, through contrasting formality with informality, narrated an additional arena where speculative work was accomplished. This was recognised by all of the team, although their perspectives differed on the accessibility of this important resource to them personally.

> *Oh yes. Doing deals. More important than giving the talks I should think. Because you can get the information from the talks anyway and it'll be published. But you can chat to people and if you get on with them get something from them which you wouldn't otherwise have done. It's called networking these days, just essentially talking to people. Figuring out who you like, who you don't like. Who you trust. Or not.*
>
> *Andrew*

The conference as an arena for potential collaboration highlighted the importance of social skills and communication. The temporal context of Andrew's description also contrasted the past with the present in the way it explored the dynamics that constituted networking. The past was conveyed as a more straightforward place where labels suggesting management were unnecessary. At the conference decisions were made regarding disclosure of information and who to trust. The scientists reconstructed the characteristics of an ideal scientist, the difficulties posed by the nature of conferences, the way management of conferences influenced their outcome, and the central role of ideas and problems as intellectual capital.

Ideal types

> *People like Tom and Bill, they're never more comfortable than with a pint in their hand and just sitting and relaxing and chatting and just mulling things and you're not under pressure at that point. You're not performing at that point. You're not presenting to people so it doesn't matter if it's a sort of really wacky idea or a really wacky thought you know, cause somebody will say "We've got a bit of this, a bit of that", and that's where your collaborations start up as well. Somebody comes up after a meeting and says "Oh, nice talk. I've got such and such. Do you think that would help you or you could help us?"*
>
> *Kay*

The narration of this ideal type emphasised relaxation and receptivity, captured in the description of the informality of the setting. The importance of this lay in the stress placed on time to "mull things over" and the possibility that through a social exchange an interesting idea might be introduced. This awareness of intellectual capital and being open to all possibilities was a theme throughout the interviews. The good idea could subvert hierarchy and convention. Gilbert and Mulkay (1984, p. 14) refer to the way "the informal talk whereby actions and beliefs are constituted at the laboratory bench is not regarded as having primacy over any subsequent interpretation around a coffee table, at a conference, in a research paper, or at an interview". This informal talk could occur and was encouraged in a variety of settings, in much the same way as the infrastructure of the team was loosely defined and managed to allow for the emergence of a good idea from any member of the team. The accounts of the DM scientists' illustrate how informality was actively promoted to enable any situation in which scientists interacted to become a potential source of information sharing. Conversely it introduced vigilance into the social

aspect of the conference where even when the scientist wasn't "performing" they were actively engaged in networking and listening for potential developments.

Being male made it easier to fulfil the role of the ideal type of scientist at conferences and meetings, according to the accounts of both female scientists. Pauline spoke of how a woman coming to the bar by herself at conferences might be misconstrued as being "there for other reasons". Negotiating unspoken agendas was an additional aspect to being a woman and militated against being relaxed and comfortable in those settings crucial for extending collaboration and investigating competition. In her account Pauline recognised that, in the years that had elapsed since gene discovery, "it may be easier...because the gender balance has shifted a bit but...A lot of it is chance." Here again the past is contrasted with the present but the outcome is uncertain and aspects of management such as gender remain subject to chance. For Kay, the technician, the conference was a place where other more senior scientists went and she stayed back because "somebody's got to run the base".

Competition and collaboration

Dynamics of competition and collaboration animated stories of scientific progress and illustrated the characteristics of good and bad scientists. This aspect of scientific work has been extensively researched in studies of the sociology of scientific knowledge (Gilbert and Mulkay 1984; Atkinson et al. 1998; Gieryn 1999). Atkinson et al. (1998, p. 261) describe groups of scientists as "characterized by movement and changing boundaries that are defined and redefined to incorporate researchers from other networks or specialities or to take advantage of new opportunities". A key turning point for collaboration related to the pooling of resources with another team when developing DNA technology could exploit the clinical knowledge of DM gained through extensive work with families.

The code of behaviour within the world of DM scientific research related primarily to trust. Contrastive rhetoric and moral accounts illustrated the ongoing adjustments necessary as the research developed and the pressure on relationships became more intense. The contingent and informal nature of the DM collaboration was conveyed in the form of anecdotes by Andrew, the senior scientist, and was accounted for by

the fact that "You should have it clear in the first place...but then people's recollection of it can become remarkably not what you thought it was." As with creativity the importance of flexibility was illustrated in justifying statements such as "It's not written in stone and it's not been run past lawyers" Accounts of justification were used to narrate strategies for negotiating the balance between collaboration and competition. They gave insight into an environment where there were ongoing shifts depending on how progress was being made and where informal arrangements were liable to change.

> *Especially I remember we had one to do when somebody was meant to provide us with some results which they were working on for entirely different reasons...So they said yes, we could have this thing to test for it in terms of DM and then they said we couldn't because they suddenly got interested in it... So we got around that by bypassing the original arrangement rather than giving in to unreasonable demands.*
>
> *Andrew*

The scientists used anecdotes, maxims and moral tales of lessons learned to give a contemporary perspective on events that had taken place many years before. The immediacy of the language and the availability of the stories within a narrative repertoire suggest the importance of these dynamics and their impact on subsequent interpretation of DM gene discovery.

For Prof the conference or scientific meeting was an opportunity to be actively managed in order to maximise benefit. Disclosure was central and necessitated "an informal, pleasant, relaxed social setting" to "get a lot of people to open up". Anecdotes demonstrated how setting an example of sharing information led to others sharing and "just set it in motion". The way that "even in the collaborations things tended to be more relaxed or tense" depending on overall progress added further complexity to the dynamics.

The competitive aspect to research groups and the way this contradicts the values of collaboration and trust has been previously noted (Traweek 1988; Atkinson et al. 1998). Prof's account further highlighted the ongoing dynamics. The benefit "of sharing and collaborating as fully as possible so everybody would gain rather than just one person" conveyed awareness of the importance of major discoveries for scientific careers. However there was awareness of competition even within collaborations, and success was "probably more by luck than judgement".

The metaphor of the "cake" and having "a fair slice" communicated gene discovery as a reward for which there would be material gain in the form of recognition in the scientific community and impact on career. Anecdotes of the danger of secrecy were constructed as moral tales where the behaviour of a good scientist sharing information was contrasted with a secretive scientist. The accounts of the DM scientists were more ambivalent and included anecdotes where being too trusting had a negative outcome. This could happen within a collaboration and resulted in uneasy tensions and potentially false information. Prof narrated how "collaboration within the collaborations was fairly straightforward and then what would happen would be that suddenly things would start to go really well and one group would perhaps find something and then it would be much more difficult to get that group...to share all that they had found with the others." The metaphor of "a level playing field" conveyed the changing nature of the terrain where "at the background was people's credit...and prestige".

The varying responses to the progress of other teams were noted in the 1998 work of Atkinson et al. The contemporary accounts incorporated potentially contradictory explanations of the significance of being the successful team making the discovery and the recognition that "You don't write on the CV 'I did this before anyone else did.' You just write I did this. I was part of this" or "The rest of the world doesn't really care who got there first. They might care that somebody got there. And that it was true." These further refined the empiricist and contingent repertoires of Gilbert and Mulkay to distinguish between the inevitable fact of the gene discovery, and the level of personal involvement. This was made explicit by Kay who explained, "There's the outside world and then there's the world where everyone knows what's going on. But then there's not that many people in the outside world that it makes any difference to in all honesty. It has to be something really phenomenal to reach into the outside world..." The way two potentially contradictory statements were reconciled through acceptance that the gene discovery was inevitable was described by Gilbert and Mulkay (1984, cited in Coffey and Atkinson 1996, p. 102) as the "Truth Will Out Device". The facts of the DM gene discovery as conveyed in the published scientific articles constituted the empiricist repertoire of conveying science as objective and value free. The context dependent nature of the

interpretation of gene discovery for the scientists was conveyed more subtly by their use of the contingency repertoire to perform a dual function of communicating personal meaning whilst recognising that there was a wider context in which the individual contributions to discovery were less significant.

Anecdotes of people who made the wrong decision or exaggerated claims resulting in their exclusion from the final success illustrated the contingent nature of success. The code of conduct was implied through moral accounts and lessons about behaviour, stressing the need for findings to be validated by the scientific community.

> *You go the meetings and people say," look at this stunning result, haven't you checked it." And then it's all degraded. And it was the wrong kind of tubes. You might go "yes I think this really is a major scientific discovery", where in fact it's an artefact which can happen with all sorts of things.*
>
> *Kay*

The regulation of scientific findings by peers within the scientific community entailed careful checking of results. The cautionary tale of the sort of situation that could arise if false claims were made, resulting in damage to reputation, highlighted the importance of the scientific community as arbiters of knowledge claims. Fujimura (1988, p. 262) notes that "scientific problem-solving and fact-making are collective enterprises" and the public humiliation of asserting a false claim was clearly outlined. Discovery as a "social phenomenon that must become an accepted part of the scientific culture to be accorded legitimacy" (Batchelor et al. 1996, p. 228) is clearly evoked through Kay's narration of a social space within which the scientist presents and is judged by peers.

Luck and judgement

The juxtaposition of luck, judgement and hard work were common to the accounts and were narrated according to the individual perspective within the team. The significance of luck for Prof was apparent in the beginnings of the DM work, in the fact that there wasn't much interest in researching DM at that stage, and also in unforeseen circumstances.

> *And then we had our stroke of luck...Which was this very nice guy called...who's an oil millionaire in Denver, developed myotonic dystrophy and had several - not congenital, but quite badly affected children. All kinds of behavioural and other problems who'd been misdiagnosed as - I'm not*

quite sure what. And eventually they found these children all had myotonic dystrophy. And I think it was only then they realised he had. And they were very devastated by this. And he asked a colleague in Denver, "Why don't you try and find people who are working on this" - because he was a very wealthy person, "and fund some projects."

Luck was related to opportunity but there was also a critical balance between hard work and judgement. This applied to funding, the work itself, the circumstances around making the final discovery and the attribution of recognition afterwards. Working hard was not itself a guarantee, as strategy and resources were also factors that could determine success.

...and probably it was more by luck than judgement, I think, that at the end of the day all the groups involved ended up with a reasonably fair amount of the credit.

Prof

It's both. It's hard work and luck. It's got to be. It's a combination of both. If you don't put the work in you're never going to get there. Unless you sit at the top and cherry pick. And wait and wait. A bit like e-bay auctioning... You wait and wait. See what happens. See what happens. And if you've got the team around you you've got everything in place and then just pull something out at the end.

Kay

The satisfaction of being successful in a field where other groups had potentially more resources at their disposal was related through David and Goliath stories where finding the gene was compared to looking "for a needle in a haystack". The metaphor of the field gave physical reality to intellectual terrain.

It still happens now. In that you have a little corner of the field and you don't want a bigger group who have more finance getting hold of something which kept you a step ahead. And them going "Of course" and ploughing all their resources into it. And getting the result before you do.

Kay

For the scientists the management of people and opportunity was not actively structured and luck could work in both directions, depriving people of recognition for years of hard work "if you come second" or conversely being able to "take that final bit" and achieve success despite being involved for a short time. Being second was the same as being last. Success was measured in publication and ranking on the final published papers which is "the bit that goes out into the world". The possibility of getting "stung" by disclosing to the wrong person or trusting someone who "lets it out of the bag" were reminders of what was at stake at a phase of

research where the goal was specific. The importance of personality in being successful, "if there's two of you racing...depends on how driven they are" again drew on the personal skills and character of the scientist in addition to intellect, hard work and luck.

The moment of discovery

The stress placed on relationships increased in the final stages. Colourful metaphors indicate discovery as dynamic and in motion.

> *They [group outside the collaboration] followed a lot of red herrings. They went up a lot of garden paths. And pursued a lot of wild geese. None of them were the right one. They had a bit of a track record for doing that so they were kind of not in at the kill...We just thought they were a bunch of cowboys.*
>
> *Andrew*

The scientific accounts constructed gene discovery as a "race" or "roller coaster". There were subtle variations in memories of the actual event, and Prof discussed the difficulty of defining the exact moment of discovery, reflecting, "Well, it's very, very difficult actually. I take the view that actually it is impossible to give a date for discovery and also impossible to give total credit to any one group or individual."

The finding of the triplet repeat, the molecular mechanism that explained the clinical phenomenon of anticipation, was acknowledged as a moment of discovery and described by Kay, the technician, as being a "bit hairy". Again she highlighted her position within the team through her response of "Best to stand back and let the bomb go off." The explosive metaphor, with its implicit notion of damage, also conveys release and detachment. The moment of clarity quickly moved to one of validating and publishing the discovery.

Reputation was linked to the ability to distinguish between a range of possibilities and clues, resulting in the successful outcome. None of this was apparent in the published papers, and the scientific world with its own norms and codes of behaviour was narrated as distinct from the everyday world. The use of the empiricist repertoire to convey the DM gene discovery in the formal scientific literature resulted in the "conventionally impersonal manner" (Mulkay et al. 1983, p. 197) of discourse where human agency was reduced to a minimum. Within the

scientific world "everyone knows what's going on" if the code of conduct was broken but there was also the recognition that "there's not that many people in the outside world that it [gene discovery] makes any difference to in all honesty".

> *I think the main thing is that it's never as clear-cut as reading papers or even hearing talks would imply. Because all the problems tend to get pruned out...if it's a paper then the methods section ought to tell people how it was done but it certainly won't tell anybody about all the false leads...*
>
> *Prof*

Use of metaphor such as "pruned" reinforced the hands-on imagery used particularly by Prof. Validating the DM discovery had implications for scientific careers and also for institutional development. The retrospective narration of events re-institutes the people involved, and the social processes by which successful gene isolation was achieved. In giving agency to the scientists to reconstruct their own accounts of gene discovery both the event and the workplace are animated and the day-to-day practices that constitute scientific work are illuminated.

Boundaries

The fascination of scientific discovery and the privilege of working with families unified a lifetime of work crossing the boundaries of different representations of DM. Prof conveyed the validity of these experiences in the form of a lesson, exemplified by the use of "one" rather than the personal pronoun.

> *Just to say that for me it's been both very fascinating and a privilege to work at this series of interfaces. Interfaces between the clinical work and the lab. The interface between research and service. Interface between the ethical issues coming out of practice and the more general philosophical and social views on them. I actually do think that there is a real value for people working at the interface. It means that quite often one isn't going to get the right answers and may limit what you can do but unless you have people at these interfaces then the issues never really get brought up and followed through in the same way and it's just incredibly fascinating and rewarding to have had all these issues arising over thirty years...and to have been part of it.*

Moral tales

The scientific team also narrated the crossing of boundaries, and in so doing they reanimated actors who remained largely invisible in the final published account. These junior doctors, who mostly progressed to careers as clinical geneticists around the UK, were invoked by reference primarily to adjusting to their presence in the laboratory. They were visible in the DM medical records through the traces of their

research projects, both failed and successful. In the narratives they functioned as an exemplar of the differences between the scientific and medical career and were remembered with varying degrees of tolerance. Kay shaped her account into a moral tale showing how the junior doctors who accepted help rather than presumed they knew it all were preferable. Images of "being all fingers and thumbs" conveyed the inexperience of these transient figures who, according to Andrew, were tolerated because "it was all part of the deal" but who also aroused jealousy because they earned more than the people who were helping them in the lab. Only Kay alluded to their wider roles noting, "often for the clinician it's a double whammy...learning what is going on and trying to juggle the lab work against the clinical side of things."

For one of the junior doctors working in the right place at the right time led to being recorded in the publication of the gene discovery. Pauline observed that "it was just luck for them that they were there at that time and they got quite a lot out of it and, you know, we'd done all the work but they got the credit and were happy to take credit for it."

Narrating these actors made them visible and they functioned to illustrate moral accounts of good and bad scientific practice, occupational differences between science and medicine, and conveyors of meaning across the boundaries of the home, the lab and the clinic. For Kay "...it was toleration because clinicians come in and they have two attitudes. One is they're prepared to learn and the other is they're high and mighty"

None of these doctors were interviewed for the project so they functioned as actors within the scientists' interpretation and memory of events rather than their own. They were evoked in every interview as essential links in a process but also personalities who affected the day-to-day work depending on their attitude to the laboratory work. In this way they served as characters to illustrate an aspect of the process of gene discovery that is no longer apparent. Although their construction as actors enabled comparison between the work of the scientist and the work of the clinician to be made, and also illustrated the contingent nature of success, the overall value of the combination of clinicians and scientists in the team was recognised. The junior doctors, under Prof's supervision, embodied this translational and relationship work. Information travelling between the clinic and the home was characterised by issues

relating to the everyday manifestations of the condition and its prognosis, whereas information travelling in the opposite direction to the laboratory was in the form of blood samples and information from research projects. Tom recalled how "they [junior doctors] were the person who really had much more of the hands-on link between the family and the sample." The imagery of hands again conveyed the DM research team as physically as well as intellectually engaged with DM in the different sites of exploration.

Hope and Hype

Interaction with the media at the time of isolation was a "bit strange actually. It was quite exciting..." according to Andrew. For him the success of the encounter with newspaper and television journalists was "because we were allowed to say it", rather than having words distorted by a journalist. The possibility of misinterpretation was extended and elaborated upon by Prof and was again characterised by a broad perspective resulting in a cautious approach. This was narrated through moral tales illustrating the consequences of naïve involvement with people who held a different agenda towards publicity.

> *I had a fair amount of contact with the media over the years. But I've always tended to avoid it on anything that's sensational or controversial which is always what they want to highlight...well, it's partly the sensational and undesirable and worrying things. Then at the same time there's great breakthroughs which are overplayed, all this. I tend to steer around the media a bit. They have been helpful at times if you get somebody with some insight. But most of the time they are just either on the scare stories or the breakthroughs which are very inaccurate. So, I tended to be fairly wary of them, actually.*

The choice of "steer" and the importance of insight gained through experience imply a career trajectory constructed around a sense of personal responsibility. This did not necessarily mean that others shared this code of conduct. Prof returned to the motivation of wanting to help families as a guiding principle. Contrastive rhetoric and examples of unacceptable behaviour again illustrated his point.

> *...it's important not to exaggerate. That's the main danger at present...especially because quite a few of those people are famous scientists maybe who've made a discovery and then say it's going to change this, that and everything else. The chances are that it won't do that but it will probably be important. People are their own worst enemies by hyping things and causing a lot of problems for more responsible folk.*

The everyday scientific work took place in a highly sociable environment and this motivation to work with people was expressed positively. The boundary between the scientists and families with DM was narrated in personal terms and was linked with a motivation to contribute to helping others, although this was pragmatically expressed. The metaphor of "divide" illustrated this boundary between emotion and work.

> *It's only a sense of curiosity when perhaps you see dates of birth or something and you go, oh, they're younger than I am or they're older than I am. Somebody to whom you associate. They're older than my parents or they're younger than me and they've got this disease...And you could ponder that a bit. But that doesn't make you good at what you're doing...you are too emotional about it then you couldn't do this job that well. If you were too emotive about it, so I think you need that divide between, you know, the people who deal with patients don't generally do the techniques.*
>
> *Kay*

> *Except as someone's said no-one's actually cured anything with genetics yet... I quite like to think that some people will get something out of it and do something good with it some day.*
>
> *Andrew*

For Prof the boundaries between patient and family organisations, and clinical research were complex, unlike the more simply expressed motivation of wanting to help families. Gene discovery as a unifying focus where "patients and patients' societies were pretty clear that finding these genes was going to be important" was straightforward, although expectations had to be carefully managed as "some of our colleagues were rather hyping it". The importance of personality again emerged with a story illustrating the good qualities of the person leading the MDSG as an exemplar for good relationships.

The MDSG functioned to contrast ideal with less ideal situations. Politics and personalities were factors in more complicated relationships with some lay groups, as was the difficulty that "Lay societies were often regarding clinics like us as a bit of a recruiting ground. And we had to kind of shelter people from the sort of overenthusiastic people who felt that everybody must join." By contrasting the role of the clinic as a "shelter" rather than a "recruiting ground" Prof also implicitly constructed the role of the clinic staff as benign and caring. The boundaries between the clinician and the family before gene discovery extended the site of research to the family home.

...it's something which I think, of all the parts of working as a medical geneticist, I think it's probably the most rewarding actually. Because it's a privilege, I mean, people let you into their house. The nice thing is that almost invariably they really are appreciative. And even though you may have gone to them with the primary aim of say getting a blood sample for research, time and again you find that you're the first person who's actually been prepared to spend time and listen. And they value that hugely.

This duty of care was recurrent through the narrative accounts given by Prof and linked the motivation to research DM with clinical commitment to the families with DM.

Lags and Phases

The major focus on DM isolation, from development of recombinant DNA to gene discovery "took ten years and so there were periods of optimism and advance in between periods of frustration." The implications of these different phases of research on the scientists were narrated across the accounts in a way that reflected science itself as a narrative. The subsequent change in focus from isolating single genes to looking at their function has resulted in changes to the nature of genetic research.

It has actually gone back to being bits of work, a lot of them quite different from one another, and so you can't say who found the function of the gene and so on. And a number of people found that very much more relaxing because there was a lot of stress in the build up to the gene and once that happened then it was back to, well, everybody's got a huge amount of work to do now to find out what on earth it does. It was much more cumulative and collaborative.

Prof

The scientists narrated their roles as actors in a bigger drama where the legacy of gene discovery led to them having to redefine themselves or become a different type of scientist as the career of science itself progressed.

when you get to the end of the genetics what are you going to do?...Either you do more genetics on another disease, or you turn into a different kind of scientist working on the same disease

Andrew

In previous research with the DM scientists Batchelor et al. (1996, p. 238, p. 245) observed, "A research group is not a fixed entity" and predicted, "This may involve the scientists' retooling or regrouping." This was borne out in the experiences of the DM team in the years following their scientific breakthrough. Narrative analysis

allowed the scientists to articulate this process from their perspective as key actors. However the legacy of their contribution to science and the validity of their contribution to genetics remained as an episode in this larger unfolding drama where science remained central. The language of mastering an elemental force was narrated as being time limited and the scientists as temporary actors in a narrative that had an infinite motion of its own.

Personal meaning

Biography

Prof narrated a moral account that moved between the personal and the impersonal as he constructed himself as a teacher and a person motivated jointly by curiosity and a desire to help. Using Labov (1997) and Cortazzi's (1993, 1999) analysis of structure it was possible to see how the gene discovery was framed within a life story or chronicle where the past is given meaning in the light of the present. Prof constructed an account of a career in medical genetics where gene discovery was significant in terms of scientific advance and in informing a personal and professional philosophy. This was accomplished through narratives of his early years as a geneticist and conveyed a picture of a personal career and the medical speciality of clinical genetics in a formative phase.

> *And two things which were relevant for later, also came up at that point. The first was the social side and seeing these people in their homes made me realise that most of them were actually very poor and living in pretty dire circumstances. And this was something which was – actually made a big impression, particularly the families living in rural America which you don't really see much of. You don't realise how much poverty there is in these little villages. And they were living in very difficult circumstances. So that was the first lesson.*
> *And the second was that I saw a lot of childhood Myotonic dystrophy. And I met up, in the course of the study with a paediatric neurologist...who'd also been interested in congenital Myotonic dystrophy. And he had noticed in his series that most of them seemed to be passed through the maternal line. So in fact we got together and realised by that time we had a rather extensive series and indeed they were virtually all maternal.*
> *And so we published that. And that laid the foundations for the congenital Myotonic dystrophy work.*

The translation of DM from its embodiment in the lives of the small town family members to an "extensive series" demonstrated the context dependent interpretation of DM, and the transformation of the family member into a case study. The location conveys DM research as pioneering and based in an unknown geography as well as

an unknown genetic map. The move to start a new department of medical genetics in the UK allowed the "foundations" gained by Prof as a training doctor to become part of his DM work as a clinical geneticist.

The linking of social situation with a genetic condition, a neurologist with a geneticist, and research grounded in practice gave the narrative context for the subsequent work that resulted in the discovery of the DM gene three decades later. Prof and the neurologist were part of the generation of doctors trained in the era that continued to recognise the clinical existence of anticipation and now placed their observation into the context of modern clinical genetic research (Friedman 2008).

Prof constructed a narrative shaped by a lifetime perspective that evaluated service and research in the context of a personal career and the impact on families with DM. He reflected on how "...it's just incredibly fascinating and rewarding to have all these issues arising over 30 years...and to have been a part of it."

The fascination of clinical work was given equal significance with scientific discovery. Personal meaning encompassed a broad perspective spanning a career defined to a large extent by DM. The significance to personal biography was repeated in the final sentence referring to the privilege of seeing through a major project in his lifetime. An additional coda reframed gene discovery as not an ending but "... a whole new start", reinforcing the image of a life cycle and lifetime perspective. The story did not end with gene discovery, which was seen as the start of a new chapter in scientific and medical advance. Instead the conclusion was offered in terms of a personal journey: the fact that "one had seen it through" and was "in there at the finish". The modest presentation of self, apparent throughout, was witnessed in the concluding remark that "You're lucky enough if you do that for one [gene]...well, that's really a great privilege".

Evaluation was also placed in the context of developing the Institute from a small department. Metaphors such as sowing reaping and ploughing conveyed skills of nurture and development. Prof described the development of the Institute through the nurturing of the skills and interests of the staff and how "... each bit turned into its own field. And so...everybody could take a bit on and it grew that way."

Ethics

Ethical issues emerging out of this developing knowledge resulted in "making policy on the hoof", and were instrumental in making a clear delineation between research and service. Once the gene was discovered this took place in the laboratory under Prof's guidance.

> *And that again was where a clinician working with the lab folk had I think an important role in this...And quite a few places that didn't do that got into deep water through coming up with finding mutations in people who, you know, had previously been told, 'Well, you've got a very low risk'...But we very much took the view that if one comes across anything unexpected one keeps it in the lab and then we evolved the situation where the service testing was separated from your research testing. It's not perfect but we never hit many problems.*

Prof then illustrated this moral lesson by telling a story about a difficult situation that had arisen. This reinforced the perspective of the moral account and of lessons being passed on through experience, again moving away from use of the personal pronoun to communicate the educational and moral aspect. The cascade effect, where being "kind of attuned to all these difficulties" in one condition also led to being able to apply them to other conditions, indicated the wider framework of looking at the implications of scientific progress.

The establishment of a reputation for expertise in DM crossed the boundaries of research and service, and as "people got to know we were interested...we set up the muscle clinic – not specifically for myotonic dystrophy, and that attracted patients". The active language of getting blood samples and visiting families at home contrasted with the way families "attached themselves to us because there was nobody else to be attached to...So we sort of served as...what you might call the general focus for these people." Being able to help coordinate management of a poorly understood condition referred back to the earlier observation of how medical management of families had been neglected. The establishment of a specialist service through a rising research profile highlighted the importance of research to institutional development and how through research the service available to families increased. As the DM research profile declined after gene discovery there was also a gradual decline in resources available for DM service.

The incorporation of the gene test into clinical service marked a definitive outcome to a particular phase of research, but the aftermath of gene discovery on the boundaries between research and service was more difficult to decipher. This is perhaps reflected in the choice of the passive tense to describe the relationship between the families and the DM clinic.

Personal responsibility for actively shaping clinical and research practice took the form of instructive accounts of justification for the development of an institutional ideology again grounded in experience and clinical practice.

> *I think we were unusual in two ways in medical genetics centres. Firstly being very, very aware of these ethical issues and also wanting to analyse. I mean, others I think were aware but they looked on them as a problem to get round, whereas we were looking on them as really a challenge to kind of put them on the agenda for the world at large...And so I think we actually did play a big role in helping to bring the kind of social science folk and the geneticists together. And I'm sure it made life more productive for the social science workers because it meant they could work on real life problems. And it was helpful for us because we couldn't take it beyond a certain point and we might have done it all the wrong way.*

The link between Prof as a motivated clinical geneticist taking a particular interest in a condition, the "lags" and "phases" of scientific research, and the changing nature of the scientific work itself had implications for families, as did the outcome of the research. Extending the ethical issues that had arisen over thirty years of DM research and service to the wider social sphere reflected another boundary or interface where the impact of genetic discovery could be analysed. Prof again emphasised the importance of grounding this in issues arising from experience rather than theory. The potential for misunderstanding and conflict arose specifically from the danger of basing research in a theoretical context without awareness of "the reality and the practice."

> *I think a lot of social scientists still, at least the ones who don't have contact with people in the field are really very naïve in what they think geneticists do. I still think they have this picture of people going around telling people what to do. I suppose if you go back to the era of eugenics that might have been the case but that's not the case now. And most families like I say are actually very realistic and very positive.*

The construction of evaluative accounts used the passive rather than the active tense to narrate how Prof responded to families' concerns. The importance was highlighted of not "going in offering wonderful new things, you know, without knowing whether

they wanted it or had heard of it. We were responding...it wasn't really very different from seeing people in the clinic or the home at the beginning. It just extended the range of what you could do hugely." The space for evaluation of the impact of gene discovery was narrated as "something which is happening and needs thinking about in order to help and not harm the people where it is arising." In evaluating the impact of DM gene discovery Prof constructed accounts that emphasised the importance that this evaluation relate to the experiences of families. The personal meaning of gene discovery and Prof's lifetime perspective returned to the original introduction of the DM work as rooted in clinical work with families.

The reference to eugenics and the dangers of "telling people what to do" were reflected in the repeated use of the passive voice to narrate involvement with families. The validity of actively engaging with the practicalities of research contrasted with the need to respond to the issues as presented by families. Use of the passive voice communicated the values of the new genetics, which lay in awareness of the past combined with the need to evaluate future developments

The Aftermath

The personal meaning of gene discovery for the scientists related to relationships with others, in addition to the impact on career and the contribution to a meta-narrative of scientific discovery. Batchelor et al. (1996, p. 248) noted the "plurality of perspectives on the breakthrough" in the immediate aftermath of the DM gene discovery. The contemporary DM accounts reconstructed conflicting emotions and ambivalence towards some aspects of gene isolation.

The scientific team universally narrated the immediate aftermath as characterised by tiredness and difficulty in focusing on the next stage of laboratory work.

> *...it is seriously like having a baby for the first time. You're only focused on delivering You don't see beyond that and that's exactly what happens with genes. People are so focused on finding the gene, finding the gene. Oh, we found it, what now. And everybody floundered for about a year, year and a half.*
>
> *Kay*

Kay compared gene discovery with "riding a wave" and again used the maternal imagery of the project as a baby. Through the use of metaphor she evoked a sense of

motion leading up to the isolation, with the resulting impact causing imbalance and blockage represented by the future as "a total closed door" where "Everybody was looking at the door and hadn't any idea about what happens next." The imagery of the sea was also used by Andrew, the senior scientist, to describe the aftermath and how "We cast around for something worthwhile and I don't think we ever got our heads around that". His account differed to Kay's in relation to both management and perception of the nature of scientific discovery.

> *...because you don't know in advance. You couldn't have done anything in advance. You don't know what it's going to be and how people are going to react so it's actually very hard to decide how you're going to deal with it when you don't know what it is.*

For Kay this lack of focus resulted in her leaving. Her moral account of the inner motivation necessary for her to carry out her job was embedded in an account of everyday routine within the laboratory.

> *Because I function at a technical level my satisfaction comes from knowing I've done the job well. Whatever it is. If it's taking the bags out to the skip. If I've done that well I'll get the satisfaction out of that.*

The metaphor of play, used earlier to construct the creative and resourceful character of the research scientist, was used differently by Kay to describe her frustration in the immediate aftermath of gene discovery where "I wasn't being kind of managed properly and I wasn't focused in what I was doing...It was well 'go and play with that' almost. I mean it wasn't like that. But it was that sort of sense of we just keep doing this for..." The value, expressed by Bill, for the research scientist of being in an environment where being "Kind of like a painter with unlimited paints and canvases with no question that what you were doing was worthwhile" was not equally relevant for the technician. The differing accounts drew out the complexity of the day-to-day work and the value associated with it. Publishing the discovery was essential for the careers of the scientists and their work was recognised in prestigious scientific journals. Kay recognised the importance of these publications for the scientific career but expressed her personal motivation through the completion of everyday tasks carried out in the laboratory. Recognition of her contribution took a different form.

The personal meaning of gene discovery was narrated some years later when Kay's work contributing to gene isolation was recognised. The integration of her personal

philosophy and the meaning of gene discovery were again expressed through the lexicon of parenthood and highlighted a different perspective within the team.

> *...basically he [DM scientist] was publicly acknowledging the sort of work I'd been doing...And for me that was like "That's my star, that people appreciate what you're doing. The actual finding the gene I would say made really little difference to my career..."*

The imagery of the "star" encapsulated motivation and personal meaning and the way it allowed her to make sense of the legacy of gene discovery within her own personal and career biography.

Registering emotion

The reconstruction of gene discovery was significant for the emotional vocabulary embedded in the narrative accounts. This register of emotion extended the function of the empiricist and contingent repertoires. The scientists acknowledged the importance of the DM gene discovery for scientific progress and narrated the contingencies that had impacted on progress, but in addition they acknowledged the emotional investment integral to their work through legacy stories of relationships. Conflicting emotions highlighted the complexity of the work environment and the personal meaning of the discovery many years later.

Pauline returned to an earlier unresolved theme of the significance of personality and gender to scientific success. The scientists were unequivocal about the scientific achievement itself. The importance of making a contribution to science was celebrated. Pauline and Prof both recognised the importance of being part of a discovery that "moved things forward". Pauline expressed the importance of "having a reason to do things" but was unsure as to whether this was "a gender thing or whether it's just me." However her ambivalence over the process of discovery and the need to be "quite selfish" was part of an account of justification for leaving the field of genetic research.

> *And I suppose the work I'm doing now is really stimulating but you know, what's it all for...it doesn't grab my heart and soul in the way that genetics did but I couldn't go back to doing genetics really. I miss it and I enjoy reading about it...but I really couldn't.*

The contrast between the importance of the discovery professionally and the implications of it personally contained the paradox of recognising the unique era and project, but also that the consequences of this made it impossible to maintain a career

in this field. The contrastive rhetoric and imagery, moving between the past and the present, conveyed the emotional intensity of the time. Reflection on the continuing importance of genetics, filtered through the activity of reading, was tinged with regret but also an emphasis on the finality of leaving the research arena.

For Prof and the scientists, even where personally ambivalent about the gene discovery, the legacy of being part of this contribution to the narrative of science was lasting and meaningful. Andrew expressed how "in other bits of science there's a whole lot of things which are not so important... Which can be superseded by better explanation. But in genetics the basics are there and they're not going to change because that's how it works. That's the way it is..." The belief in the importance of genetics as a paradigm of fundamental importance was emphasised and was again narrated as being primary in a hierarchy of knowledge.

The theme of legacy emerged in relation to genetics in science and the role of the scientist as teacher. This served the dual function of communicating a scientific event of consequence, and of keeping it alive in scientific culture by passing it to future generations. The relationships and cooperation integral to the process of scientific discovery were recognised and emphasised. Bill reflected how "I have never mixed with a brighter bunch of people and have subsequently been looking to help generate the same type of environment." Several of the scientists, now in senior roles, described how they tried to replicate this environment for their junior staff.

The importance of mentoring and encouraging the career of younger scientists was part of this legacy and combined both practical and aspirational aspects. The emphasis reappeared of the quality of the ideas and personnel being more important than resources. This, together with the focus on less tangible outcomes, such as fulfilling not just expectations but "dreams", and making quality relationships, presented an additional narrative of scientific work to official scientific discourse.

> *I've very happy memories of working in [the Institute] which is as much to do with the people as the job. I've never really got that back in anything I've done since and I don't think I ever would.*
>
> *Andrew*

The personal meaning of gene discovery evaluated the significance of relationships and emotional experience in addition to scientific achievement. Contrastive rhetoric

compared the past with the present. The significance of gene discovery as an event fundamentally shaping personal outlook as well as relationships with others was evident. The scientists constructed narratives of a unique time that was either impossible to repeat, or an aspiration to recreate. This was evoked through language of idealism and echoed the opening of the scientific interviews, where a type of person and a way of working was narrated.

> *I also think there is still a place for what has been called the serendipitous discovery, the translocation or deletion that reveals a common phenotype. In the end there has to be a person with a theory putting it all together. I'm quite keen to continue to try to be one of them.*
>
> *Bill*

The placing of a "person with a theory" reinstated the scientist as central to scientific discovery and reinforced the narrative function of illuminating the processes and people that were part of the DM gene isolation, rather than privileging the published scientific account.

The impact of gene discovery

> *It's revolutionised things. It makes an interesting contrast. Because for probably five, almost ten years after the isolation of the gene it didn't actually help at all in understanding what it did or what was going wrong. But it made an immediate impact on what you could do genetically. I mean, first of all you could use it diagnostically, so that – there were puzzling patients who nobody knew, did they have myotonic dystrophy or is it something else? And you had a test which was absolutely clear- cut. And then for family members it meant that instead of saying to somebody who seemed to be alright, "Well, looks like you're alright, we can't find anything. But, you know, you'll have to wait a few years." And they would still be worried might they pass it on. It meant that you could say, "Well, we can make absolutely sure by offering a test." And so you could completely exclude being – carrying the gene. Whereas before you could actually go a long way but – and with the linked markers that helped too, but it wasn't the same as being absolutely confident.*
>
> *Prof*

Two of the original team, Prof and Tom, continued to work on DM after gene discovery, although Tom moved to another location and Prof's focus was primarily clinical. The condition of DM, as a focus of research interest, attracted modest interest, as noted by Andrew, who commented on the "remarkably small number of people in the rest of the world" working on it.

Gene discovery as a turning point and as a contrast between the present and the past was strongly emphasised by Prof. The strength of the comparison to revolution contrasts with the modest language used elsewhere, particularly with presentation of self. The role of the clinical geneticist in establishing certainty was evoked, as were the implications of diagnostic uncertainty in the past. The application of technology in the form of a gene test placed evaluation of gene isolation in the context of what it could offer in the clinical situation. This related to the impact on the geneticist's job, and on the family in the clinical encounter. The temporal aspect to negotiating uncertainty, particularly with regard to reproduction, changed from years in some situations to immediate certainty. The temporal aspect of research itself and reaching understanding was referred to by Prof, but it was the immediate possibility of making an impact on the clinical situation that provided the central focus of evaluation.

Tom also referred to changes in the clinical context. These were narrated with ambivalence and an anecdote from a family with DM provided a perspective that shifted evaluation from objective to subjective. Contact with families "brings it home to you, the impact on the families. It's not just a set of DNA or a bunch of cells in a lab." The temporal aspect of research, referred to by Prof, was explored with the ultimate aim of finding a treatment. Acceptance of latent and more active phases gave a temporal trajectory to the nature of research itself. Tom, like Prof, narrated a long- term perspective but this related specifically to the way scientific progress happens.

> *How have patients benefited? Well I think very much in terms that the main problem facing most inherited conditions is that you can offer pre-natal diagnosis. The outcome of that might be termination of pregnancy. We haven't got anything really positive to say. You can say, well now there's a genetic test, and in fact somebody came up to me at the last patient support group meeting and said, "I'm so grateful for your efforts. My daughter had a pregnancy. She had a pre-natal diagnosis and we knew that the baby was unaffected because of the test you'd developed and thank you so much for that." And I felt really quite humble and strange. It doesn't happen very often, that kind of thing, but obviously that's the implication of developing a genetic test. But I think, ten years might be a bit rash, but I don't know. We could really have things to help treat the condition which just would not have been imaginable fifteen years ago, but now suddenly it is and it seems to have taken a long time from gene discovery to where we are now, but a lot has been learned and we've got good insight into the molecular basis of the condition and now really it is into the next phase and often things go from, myotonic dystrophy first described around 1900 and then it takes a long time*

before DNA technology changes to find the gene and then there's another lag. But it's another phase.

Understanding of the molecular basis to anticipation answered long-standing clinical questions and enabled the development of a DM diagnostic gene test. Cooperation between families and researchers at the Institute facilitated the clinical knowledge of the natural history of DM that played a key role in the contribution of the local research team to the international collaboration. Prof illustrated the challenge of not only understanding a condition such as DM, but also understanding a person with the condition in his observation that "I think more with myotonic dystrophy than any other disease I can think of...there's this tendency to minimise, downplay, even deny a lot of problems." The construction of DM by the families interviewed located it in the everyday, rather than as an illness or disease. The classification of problems medically and genetically, and the perception of those problems by families remain an area for negotiation and the necessity of reaching shared understanding in the clinical situation.

Summary

Analysis of the accounts of gene discovery related to less tangible but nonetheless influential factors such as personality, conflict, motivation, reputation, complications and contingencies. The rhetoric of luck and judgement and use of accounting devices have been documented in previous work exploring the nature of scientific culture (Gilbert and Mulkay 1984; Atkinson et al. 1997).

Narrative analysis in this thesis drew out less recognised aspects of scientific practice such as the roles of friendship and loyalty, the challenge for scientists of narrating dynamics within scientific work, and the personal meaning of being part of a scientific discovery. Analysis indicated the use of the empiricist and contingent repertoires but also a vocabulary of emotion that distinguished the accounts of scientific discovery.

Narrative analysis provided a way of exploring the culture of scientific discovery and of opening up previously recognised phenomena such as networks of competition and collaboration to closer scrutiny. It brought to life the dynamics of what it means to be a scientist working in a novel area. DM primarily appears in scientific or

medical accounts. These accounts are characterised by a formal structure and a privileging of facts over meanings. They reinforce the discourse of science and medicine as empirical and objective. By opening up narrative the actors were placed centrally and allowed to tell their own story. By restoring agency insight was given to the process of gene discovery and the way scientific discovery was experienced by the key actors.

The DM team used narrative to reconstruct an ideal type of scientist and a unique era of scientific work. Through this they highlighted the importance of personal characteristics and a work environment conducive to creative and flexible problem solvers. The importance of an intellectual space within which management or imposed structures could not confine ambition and scope was highlighted through construction of good and bad practice. However roles within the team became apparent in the accounts that were the exception to the rule. In this way Kay illustrated the disruptive consequences of gene discovery on her work of maintaining a laboratory routine, and Pauline gave an insight into how gender was an additional factor when accessing informal but influential networks within the scientific world.

Gene discovery was framed in accounts that constructed its personal meaning and its relevance within a broader narrative of scientific progress. In doing so they privileged friendship and loyalty, while at the same time resisting a definition of the everyday dynamics that underpinned progress. Through moral tales and contrastive rhetoric they recreated the competition and collaboration that characterised scientific research and also evoked actors whose role has remained largely invisible. In this way the storying of relationships as well as results allowed the practice of both good and bad science to appear.

The analytic approaches taken demonstrate how scientists constructed narrative accounts of their careers and how scientific work was accomplished. Narratives do not operate in isolation and are peopled by other characters. These may be used to illustrate a moral tale, such as the hero and the villain, and "in most instances the actions of others contribute vitally to the events linked in narrative sequence" (Gergen 2001, p. 252). Contrastive rhetoric highlighted desirable or unacceptable behaviour while accounting devices placed actions and outcomes in their cultural

context. In the case of the scientific interviews there was a cast of characters from the core team of researchers and the wider academic and scientific world. What was notable was how the retrospective accounts were constructed to re-assemble this cast and to acknowledge the less obvious aspects of being part of a successful scientific endeavour such as friendship and mentoring.

Interview contact gave a unique opportunity for the scientists to reflect on a time of major scientific advance and how it related to their personal biographies. The gene discoveries were made during a period when the plan to map the human genome was starting and gene identification was anticipated to presage advances in the understanding, diagnosis and treatment of hereditary illness. The evaluative function of the narratives privileged relationships and networks as well as the enduring status of the gene discovery within science as embodied in the published scientific articles. The analysis revealed an approach towards scientific progress that was grounded in pragmatic hope rather than speculative hype.

The gene discovery is now embedded in the biographies of the scientists, each of whose narrative had a highly individual trajectory. While the researcher acknowledges the co-construction of interviews the narratives reflect individual agency in the choice of language made to construct them. Scientists' accounts were replete with characters offering insight into where and how scientific work is accomplished and examples of desirable and unacceptable behaviour. The narratives gave accounts of DM gene discovery but were rich in detail about the importance of the team. The legacy of friendship was apparent as much as the legacy of the effects of gene discovery on individual careers. The narratives were detailed and multi-faceted, incorporating emotional as well as intellectual responses to the memories of the time.

Chapter Five

Bound Together: Research and Service in the DM medical record

Introduction

First we went through your story. You have noticed that you are tired all the time and that you have a tendency to bump into things. You tend to fall when using steps and trip over uneven pavements. You do not drop things. You have noticed that your hands become very stiff at times...Considering your difficulties and family history, you too are now wondering as to whether you have md. When I examined you in clinic you had physical signs that I would expect in a person with md. Your story, in combination with your family history and examination suggest that you also have md. ...the genetic change in md is an expansion within the md gene. The amount the gene has enlarged is variable. The size of the enlargement is associated with the severity of the condition. Due to the limitation of the techniques that we use to look for changes in the gene, we can only say whether the gene is normal, slightly enlarged or very enlarged... I enclose the anaesthetic risk card. The address for the MDSG is on the back. We will arrange to see you again in January.

Letter from geneticist to family member following clinic 2002

The above excerpt captures DM as it is experienced in everyday life and as it is classified medically. It demonstrates the use of narrative as a way of reaching shared understanding in the DM clinic and illustrates the ongoing uncertainty about the DM phenotype; in particular the individual implications of the triplet repeat expansion. Knowledge gained over years of documenting the natural history of DM resulted in improved aspects of medical care, such as awareness of the dangers of anaesthesia in DM, and the fostering of good relations with the MDSG is evident in the inclusion of their details. The clinic as the site of interaction reflects the contemporary management of DM, and the letter addressed directly to the family member is integral to the relationship between geneticist and family from the very first DM record.

The DM medical record archive is a repository of over three decades of research and service. It functions as an intrinsic part of the medical genetic service at the Institute, a testament to a unique phase of research grounded in work between families and professionals, a history of the classification of an inherited condition, and a record of the trajectory of DM for individual family members.

This thesis did not treat the records as transparent testimony of a single story. The DM archive contained many perspectives and representations of DM. A key feature,

particularly of the early records, is communication with professionals in other areas of the country. These are referred to as "out of area" to distinguish them from staff at the Institute.

The medical genetic records or notes comprise the particular linguistic repertoire and set of practices associated with clinical genetic medicine. Their function is to record the interactions of families and professionals through their interpretation of and decision-making about DM. This implies an understanding, or at least a working definition of DM that could be mutually shared. The notes capture the way in which this understanding was communicated and negotiated or rejected through language, primarily metaphor.

They were interpreted as documents whose "production, distribution, and preservation" were integral to the "collective actions" (Hammersley and Atkinson 1995, p.157) of geneticists and families and a fundamental part of the culture of clinical genetics. Rees (1981, p. 68) describes placing the record "in the working context so that we can make sense both of the way it is constructed and the way it is read". The DM notes functioned as indicators of past activity and were instrumental in shaping future activity through iterative practice. Junior doctors learnt how to shape the clinical or home visit encounter from preceding consultations documented and formatted within the record (Pettinari 1988). The notes performed a pedagogical function and also presumed expert readership through information summarised in short histories. The origins of the DM protocol, now standardised as a form in the DM clinic, could be seen in the handwritten bullet point lists of the earliest entries.

Analysis of the DM archive showed how classification and practice were influenced by the gene discovery, but how key aspects of DM in the context of clinical genetics continue to be the complexity of managing family relationships and the uncertainty underlying DM manifestation.

The origins of research and service within the emergent paradigm of clinical genetics are visible in the medical records. They trace the evolution of the DM classification and the incremental progress towards diagnostic certainty, resulting in the development of a diagnostic DNA blood test. Traces of research projects record how

research progress was bound with service. The consideration and discarding of hypotheses, as an integral part of research progress was evident and the records documented the series of junior doctors who explored these possibilities as they acquired the specialist skills of DM management.

> *I would however be grateful if I could arrange to collect a further blood sample. There are many unsolved problems in the cause of this muscle disease and part of this project involves looking for a biochemical abnormality in the blood. This is an entirely new approach and one which we are working on with colleagues at Duke University in North Carolina...*
>
> *Junior doctor to family member 1980*

The gene discovery was significant in the wider world as part of the narrative of the new genetics. The records highlight how smaller contributions towards progress such as the re-evaluation of risk in families were modified by increased knowledge through research such as linkage analysis. Although less dramatic they indicate a trajectory of incremental knowledge leading to the development of a specialist management clinic that reached a zenith in the years around gene discovery. The intertwining of the research and service narratives was a fundamental factor in the successful contribution of the local team to the international collaboration. It was grounded in specialist clinical expertise in the natural history of DM gained through relationships with families. The way this sharing of knowledge about DM was translated through narratives grounded in everyday life influenced the research and management of DM.

Representation of DM in its everyday manifestations, in addition to its molecular and medical context, distinguished the relationships between families and clinician. Contemporary DM management is rooted in past practice but changes in practice also reflect the implications of the definitive DM diagnostic test. The collecting of information about the natural history of the condition has continued but without a specific scientific research focus at the Institute. However the success of DM gene discovery and the relationships developed with families led to the establishment of the current DM research register. Recruitment of families interested in taking part in future projects may form a key part of the next phase in DM research.

The archive is rich in potential research material and represents a significant resource. This analysis is designed to offer perspectives on major themes and implicit in this is the acceptance that scope for further research remains.

DM, the Clinic and the Family

Relationships

The observation that "plans are also material resources for action" (Bowker and Star 2000, p. 54) could equally be applicable to the notes. Once a set of notes came into being there was a template for future activity. There were two main aspects to communicating DM: firstly defining DM as an entity, and secondly dealing with DM through activities such researching it, living with it, diagnosing it or predicting a future with DM.

Defining and dealing with DM both involve perceiving DM as an active agent of change. This was presented in the first main activity of the notes in the visual metaphor of the family tree, which structures "the scene and mediates the interactions within it" (Prior 2003, p. 50). The identification of DM temporally, in motion over generations, implies its ability to move through time. Hitherto unremarkable signs and symptoms such as cataracts in a grandparent become significant in the light of a potential inherited disease. Activity was required by the family member to reconfigure their family history into a new one defined by DM. Through this work "The pedigree does not merely report or picture family relations; it is one mechanism whereby such relations – and, indeed, families themselves are produced actively for the practical purposes of clinical genetic work" (Latimer et al. 2006, p. 611). The family tree symbolises the future as well as the past and represents the reference point for the mapping of future events in the light of a new familial reconfiguration of DM.

This ontological and orientational metaphor of DM as an agent of change places the geneticist and the family in a shared frame of reference about DM as a journey or process, rather than as a diagnosis and treatment framework common to other specialities of medicine (Lakoff and Johnson 1980). There was a particular language used in the notes that reflects insider knowledge and by implication initiation through

language of new members. The notes illustrate how "specialized script can enter into, and structure, social interchange" (Giddens, cited in Prior 2003, p. 53).

> *First the name of your condition is myotonic dystrophy. The term myotonia means muscle stiffness.*
>
> *Prof to family member 1997*

The everyday was the common ground through which reinterpretation took place. Observations and memories were re-evaluated and translated into DM. Lakoff and Johnson (1980, p. 54) refer to how "conceptual metaphors are grounded in *correlations* within our experience".

> *Her arms and particularly her hands are weak and she drops things and is unable to carry such a thing as a shopping basket in her hand and has to put it over the crook of her elbow.*
>
> *Out of area physician to GP cc. Prof 1975*

The shared understanding of DM between the family and the geneticist through the metaphor of DM as a journey or agent of change was translated through the reinterpretation of family experience in the language of everyday life. However the interpretation of DM for family members was grounded in their individual experience of the condition, and in their acceptance of the shared metaphor. In families where there was a recognised history of DM reference was often made to the previous generation and how they had coped with DM. The experience and the way they dealt with it were significant for the decisions of the next generation. The metaphor of the journey was rejected in some instances leaving unresolved implications for other family members. In this way the grounding of inherited disease in the wider family, despite the availability of a gene test for individuals, could be seen.

The reconstitution of the family member into a patient who needed monitoring and surveillance implied an uneasy alliance between the trajectory of an uncertain condition and the trajectory of a person's life. A central ritual associated with the metaphor of DM as a journey is the measuring of this progress through attendance at the specialist DM clinic. The origins of the clinic lie in the foundations of research and service before gene discovery. Initial referrals for genetic counselling and diagnosis provided a basis for developing the DM protocol used in contemporary practice. This was located in assessing the situation and the issues from the family point of view, and involved introducing them in many cases to a new paradigm of

inherited disease. The earliest research, often located in family homes, introduced the representation of DM as experienced in everyday life. The combination of family history, developing clinical knowledge and awareness of how the condition affected everyday life could be traced in the observations and examinations written in the notes. The evolution of the clinic represents a validation of DM as an inherited condition and provided a place to assess the natural history of DM in addition to providing advice on management.

The notes up to gene discovery indicate the clinic as having a dual function, and being staffed by junior doctors with specific research projects as well as service commitments. In the years after gene discovery the connection between scientific research and the clinic declined and medical management predominated. The DM clinic also entailed a more defined journey through protocol and bureaucracy, where genetic counselling and diagnostic consultations were carried out before referral to the specialist clinic. The "progress" of DM continues to be "monitored" and "followed up" here through a standardised protocol closely based on the original examination. Diagnosis extended the use of the metaphor of motion in the description of its potential "impact" and anticipation of both positive and negative aspects of having a gene test are part of contemporary discourse around diagnosis.

The earliest notes indicated how the relationship of families with clinical genetics was initiated both by referral for genetic counselling and diagnosis, and by the geneticist recruiting families for research. Referral for a service reason did not preclude becoming involved in research. The embedding of research and service in the foundations of the Institute is documented through letters outlining the purpose of visits.

> *This would involve me coming to your home to take a full family history from you, a brief examination and provide you with a good opportunity to answer any question which you may have pertaining to the muscle condition.*
>
> *Geneticist to family member 1991*

As Lindee (2005, p. 201) notes:

> Virtually all scientific and press reports about newly found genes include a sentence that proposes that finding a gene will lead to a cure for the relevant disease. This is ubiquitous enough to be understood as a literary convention in genomics... The proposal that the gene will lead to a cure is the primary explicit justification for the search for disease genes...

Research itself was a journey with reference to progress and hope for the future. The implied destination was treatment and eventually a cure. Sharing of this understanding ensured continued participation of families with research.

> *In the past you kindly helped us by donating a blood sample for family studies on the disorder md. You may have heard either through us or through other sources, that we and colleagues have had success in isolating the md gene. This is bringing real progress...We are writing now because our continued research involves testing the stored samples on family members...*
>
> *Junior doctor to family member 1993*

Relationships with other clinicians, mainly in neurology and paediatrics due to the manifestations of DM, were essential to the setting up of the first major research study in the early 1970's, grounded in clinical observation and expertise, and focused on the phenomenon of anticipation. The relationship between maternal transmission and increased severity of DM underpinned the research. The work of managing relationships was primarily carried out through letters.

> *A survey is being carried out...The study is particularly aimed at identifying the genetic and other factors responsible for the early occurrence of symptoms in such patients. It is hoped that information will be carried out on all known cases in Britain*
>
> *Prof to out of area paediatrician 1973*

Communication with fellow professionals did not always result in contact with families although some details could be gained from family records. Research as a justification for renewing contact appeared as a priority even where the diagnosis was unclear. The way ethical guidelines for family research have changed (Hunter et al. 2001) in the three decades since DM research began serves to frame the pre-gene era in the notes as unique.

In most cases the notes indicate that good ongoing relationships resulted in a negotiated maintenance of contact. Notes before gene discovery reflect the informal and cooperative nature of service and research interaction. Letters from families updated the geneticist on relevant details, sought advice, and offered insight into how DM affected carers as well as diagnosed family members. A 1981 letter from the husband of a woman with DM noting that, "...you mentioned that you had written a book about md; is this a purely medical textbook? Or is it likely to be of value to the layman?" suggests deference and respect but also ease with communicating directly with Prof. The content of the letters asking advice on how best to manage DM were

related to a variety of issues from vitamins, diet and exercise to difficult family relationships.

> *I am moved to write to seek your advice concerning my wife, who is on your files as a sufferer of MD. What I am concerned with, is seeking some kind of advice and guidance about the future progress of the condition and the deterioration in my wife's health...I trust there is nothing unethical in my writing to you in this way. I stress that it is entirely without my wife's knowledge, but I feel I really must seek some guidance and I have to do so surreptitiously.*
>
> *Husband to Prof 1981*

New personnel appeared in the notes, reflecting the expanding department. Their contributions traced the process of becoming initiated into the practice and research of DM. In addition to taking family histories these junior doctors carried out examinations. Bullet points or lists related to specific signs or symptoms show how the profile of DM was elicited.

The involvement of these junior doctors with research can be seen through the letters recruiting family members. The junior doctors legitimised their work through highlighting their relationship with Prof and the continuing relationship with the families and research into DM.

> *I work with [Prof] on research into myotonic dystrophy...Since your family was last seen there have been some further developments in the diagnosis of myotonic dystrophy and this could in future lead to early detection and hopefully improve methods of treatment...*
>
> *Junior doctor to family member 1982*

It was only when communication became a problem that the underlying work involved in maintaining good relationships with families became visible. The delicate balance between the research and service agendas lay in negotiation and interpretation of ethical boundaries.

> *I write to complain in the strongest possible terms about the behaviour of Dr [junior doctor]. He induced me to bring my wife on the grounds that it would be to her personal benefit, rather than making it clear that the purpose of her visit was primarily to participate in a piece of medical research. I wonder whether you or Dr... have the faintest idea of the cost to my wife of such a visit. I am also bound to wonder what happened to the concept of medical ethics.*
>
> *Husband to Prof 1981*

Addressing the complaint to Prof signified the issue as a breach of trust rather than a misunderstanding about a separate hearing test, that was part of the research interest

of the junior doctor, being added to a clinic visit. This resulted in a sequence of letters from the junior doctor and Prof explaining the circumstances around the visit that had led to misunderstanding but also invoking altruism and helping others as a motivation for participation.

> *I am saddened that I should have caused you such inconvenience and hope that you will accept this sincere apology and allow me to try and explain.*
>
> *Junior doctor to husband 1981*

> *The reason that Dr [junior doctor] arranged the hearing tests was because we have found many patients with md having problems and in a number of cases have been able to help them directly, as a result of the tests quite apart from learning more. Neither Dr... or I would wish to put anybody to unnecessary inconvenience so we do apologise if this has been the case...*
>
> *Prof to husband 1981*

The context of research after gene discovery lay in NHS protocols standardising and bureaucratising ethical consent. The original DM study into maternal transmission could not be instigated now without nationwide application to every NHS Trust before approaching paediatricians, GP's and neurologists. Current records showed little direct involvement with scientific research. The contemporary DM clinic acts as a pathway to draw attention to research, mainly focused on the medical management and social issues related to DM. However the potential for future research continues, as indicated by the continued collection of clinical information through the DM protocol and the DM research register.

The relationship between technology and the clinic was visible in the application of tests such as EMG prior to gene discovery. The extent to which relationships between the laboratory and the clinic were integral to the DM service following gene discovery became apparent in rare instances when problems arose.

> *We have received your request for testing for md along with a sample of blood from your patient. Unfortunately the lab has already exceeded the number of investigations agreed in the contract for this financial year with the purchasers. The cause of this overactivity is an ongoing increase in the referrals made to the lab...We regret any inconvenience this may cause however this action is necessary to improve the quality and ensure the continued development of the molecular diagnostic service...*
>
> *DNA report 1997*

Relationships in the DM notes were made explicit through letters and handwritten notes, and also functioned at an implicit level visible in the standardised forms for laboratory services. Documentary analysis highlighted how key activities in the notes

were mediated through these relationships and the work that was necessary to maintain them.

Risk

Communication relating to risk was central to the DM notes. Before gene discovery this related to diagnosis and reproduction. Reproductive issues and their interpretation by other medical specialities necessitated advocacy and reframing of DM as an issue requiring information for decision-making rather than a problem itself.

> *...please use any of my cases...Fortunately the patient's daughter, although married, has not bred, although I did not enquire into the reasons for this.*
>
> *Consultant neurologist to Prof 1976*

> *When the md subject came under discussion she [wife] was advised not to have children, as the consequences could be very serious for us as in looking after handicapped children. She [gynaecologist] had not heard of the disease before.*
>
> *Husband with family history of DM to Prof 1989*

Negotiation of risk was framed around clinical examination, use of technology such as slit lamp, and application of new knowledge such as genetic linkage that altered risk status. The notes include communication directly from families, often over many years, giving insight into the stress of living with diagnostic uncertainty. Renewal of contact often coincided with a major life event such as meeting a partner or reproductive decision-making, as the extracts from a young man below indicate.

> *You may remember me as the son of the late... You examined all our family and said that if we were ever concerned about the slight chance of having this possible hereditary disease we should contact you...I would be grateful for your advice. I am still a bit clumsy and find it difficult to stop rushing things...possibly myotonic dystrophy. I would like to thank you for your care of my father...At the moment I feel in need of reassurance which I hope you can give me...*
>
> *1979*

> *I was wondering if you would be kind enough to see my wife and me about my taking the tests again for md and explaining the disease and risks involved in having children.*
>
> *1989*

Notes before gene discovery captured the stress associated with uncertainty for families and the work of the clinic in managing risk through ongoing interpretation. The description of "the course" of DM, its "progress", remaining "stable",

"following" the patient, is indicative of motion and progression. The work of the geneticist in routinising and managing uncertainty was to emphasise the slow rate of this progression, in the absence of real certainty about the future. The application of relative risk figures to DM before gene discovery framed uncertainty, where anticipated events along a trajectory of family life such as having children were reconfigured into risk narratives.

> *...has now become informative for the probe D10/Pvu2. This means that there is a 75% chance that she will be informative during a pregnancy should she wish an antenatal test to be done...*
>
> *Junior doctor to consultant paediatrician 1990*

Diagnostic certainty was established following gene discovery and the advent of the gene test but this did not necessarily make reproductive decision-making easier. Although gene discovery offered diagnostic certainty the representation of DM as unpredictable remained.

> *One slightly confusing thing about md is the nature of the gene alteration... Part of the explanation lies in the tendency for the repeat size to get bigger going down the generations and sometimes for this increase in size to be quite a big jump.*
>
> *Geneticist letter to family member 2005*

Hopes after gene isolation that exact repeat size would correlate to severity of the phenotype were unfounded. Reference to the specific number stopped in reports, although it could be invoked in exceptional circumstances by reference to Prof as the expert.

> *I therefore discussed her case with [Prof] and he felt that the first thing we would need to know is the exact size of her expansion.*
>
> *Geneticist to referring out of area paediatrician 2002*

Metaphors suggesting DM as an unpredictable and mysterious agent were common in the family interviews but were also apparent in the notes and in references to DM in medical and scientific literature. Gene discovery led to identification of an unstable triplet repeat, echoing the management metaphor of stability. "Isolating" the gene inferred tracking it down. The language of detection and surveillance was common throughout the notes with reference to "evidence", "suspicion" and "pending the outcome of these investigations". DM as "badly behaved" (McGuffin et al. 1994, p.197) reinforced the classification of the condition as highly variable and unpredictable. This aspect of DM was common across the data set before and after gene discovery, with the likening of consultations to detective narratives looking for

"clues". Allusions of deviancy necessitated "surveillance" but also "tracking down". Descriptions of DM by geneticists as something arising "out of the blue" or "which has only come to light this last six months or so" share a common metaphor of visibility with family descriptions of DM.

A handwritten letter to Prof in 1974 from the mother of adult children who "have had tests for Dystrophia Myotonica and have been passed as completely clean...I would very much like to contribute in some small way towards the research with this terrible disease" also conveys cleanliness and absolution. Similarly the metaphor used by a young woman requesting pre-natal testing in 2007 as "she wanted to wipe out this gene" conveys personal responsibility and cleanliness.

Individual and familial knowledge and experience of DM was a major factor in deciding how acceptable a risk was in reality.

> *...does not see this [DM] as a big problem, there have been no major medical problems in the family.*
>
> *Genetic nurse specialist 2004*

> *...feels that her father's condition commenced five years prior to his death when his speech became slurred, weight loss and he began to sleep a lot...However his condition is not discussed much amongst the family...*
>
> *Genetic nurse specialist 1996*

Following gene discovery protocols and standard practice for dealing with testing began to emerge. Pre-symptomatic and pre-natal testing necessitated communication about negative as well as positive consequences. Issues such as insurance implications were raised as part of a staged protocol with the option that "if at any point you decided that you wanted to stop either because of wanting to sort out other issues such as life insurance or for any other reason then that would have no impact on your option to be tested again in the future..." References to the possibility of "survivor guilt" in some letters indicate how awareness of psychological as well as physical and practical implications of testing became integrated into genetic counselling practice.

Concern for others was apparent in self-referrals to the GP for gene testing. In some cases the family member was more informed than the doctor.

...His very sensible sister is already very well aware of the genetic possibilities and what can be achieved by testing but I am simply not up to date myself and would be grateful if you could advise...

GP referral to the Institute 2005

Worries about other family members were evident particularly where younger generations were of childbearing age. The gradual introduction of pathways of referral and protocols for testing also reintroduced the home visit as the site for reflection on the issues. Clinic and home visit notes contained lists of questions asked by the family member, although recorded by the clinical geneticist or genetic nurse specialist.

What is MD. Is there a disease spectrum. How is it inherited. Can a gene test be done. What are the implications of having a gene test. What health surveillance is advised in those who have the condition. What does it mean for the girls.

Genetic nurse specialist home visit notes 2005

The notes capture the sense of responsibility attached to contacting younger generations when they reached reproductive maturity. There did not appear to be a clear protocol over how exactly this should be done. The notes serve as a reminder of work with future generations in addition to the current one.

As you are aware you have a f/h of md. This condition is inherited. It tends to get worse with each generation and the children of females are more at risk of having severely affected babies. It is possible to do pre-symptomatic testing on young women. If you would like to discuss this please don't hesitate to contact me and I will arrange for you to be seen in the local clinic.

Geneticist to sister of woman with DM 2005

Responsibility in situations where dynamics made communication with extended family difficult illustrate how technological advance such as the gene test remained dependent on personal interpretation of its function.

...has no contact with his first wife or daughter. I pointed out that because of the inheritance pattern that both his daughters are at 50% risk if he is given a diagnosis of MD. ...says he would find it difficult to contact his daughters...

Genetic nurse specialist home visit notes 2005

Classification

The notes document how classification in DM has been an active and incremental process, in which the recording of DM in the notes played a key part. Gene isolation led to diagnostic certainty but the phenomenon of anticipation continued to be emblematic of uncertainty in the association between triplet repeat size and severity

of prognosis. The unreliability of the phenotype, described by Lindee (2005), reflects the idealized and reductive nature of classification of DM.

Classification work by the families themselves was evident, particularly in letters in the early notes where identifying familial patterns fulfilled a dual function of contributing to DM research and incorporating understanding of the condition. In a handwritten letter to Prof a young woman reflected that those members of her family diagnosed with DM "never seem very happy or very sad or so it seems". Handwritten letters from family members were a feature of the decades leading to gene discovery and were primarily addressed to Prof, indicating the fluid nature of relationships in the early stages of DM research and service. Understanding what constituted DM in previous generations became part of the work of trying to make sense of a potentially altered self for the future.

The communication and classification of DM across different settings, starting with the home and the clinic, but extending to the laboratory and then to the various specialities of management, exemplified its work as a boundary object (Star and Griesemer 1989; Cambrosio and Keating 1995). The way subtle changes were made to communicate understanding in context reveals the flexible nature of classification in practice. The invisible work of classification became visible in the occasional failures where there was a refusal to accept the definition given by the geneticist.

> *Dear Madam, I write to you...severely restricted movement...actually has difficulty getting around her house...I would appreciate if you could send me a report as requested before...*
>
> *Geneticist to physiotherapist 1990*

> *Dear Dr..., This lady has had regular visits...I think she lies around most of the day as she is usually still in her dressing gown late afternoon. I do not think we can offer her any extra physio at the hospital as she is very mobile when I visit her...*
>
> *Physiotherapist reply to geneticist 1990*

The notes trace differing interpretations of classification between geneticists in different parts of the country, requiring diplomatic communication. Short social histories reflected typification and the challenge of fitting a classification of a variable condition in real life.

She coped poorly as a housewife.

Out of area geneticist to paediatrician late 1970's

She lives with her husband, drinks 10 units of alcohol a week and impressively has four jobs.

Cardiologist to geneticist 2006

The correspondence from Prof was characterised from the beginning by considered responses to potentially difficult situations. Referral of the "curious" or "interesting" cases to him reflected his status as an expert but also highlighted the necessity of maintaining good relationships.

Variations within families regarding classification could be seen where some attended clinic and others chose not to, or in one case where a father attributed an improvement in his condition to a religious cure. The visibility and plausibility of DM as a classification outside the medical setting was a source of anxiety in the family interviews and DM notes. Early correspondence highlighted the role of the geneticist as a boundary crosser. Requests such as "I would be very grateful if you could let me have some sort of official looking statement that I can show to the people concerned" highlight the communication of appropriate versions of DM in different contexts.

Later notes contain many letters written on behalf of families to funding bodies for allowances and equipment. The advocacy role included legal and insurance issues as well as practical and medical management. Responsibility for protection of genetic information was apparent from the construction of the first notes and the issue of ownership of information within the DM classification related to principle and to practice. Classification in this instance was responsive to situations rather than referential, and involved the anticipation of controversial issues where there was no previous practice to follow. Aspects of classification such as pressure to disclose genetic information to an insurance company revolved around ownership and guardianship of information. Prof's response, stating, "I would be happy to disclose information about this patient but only on receipt of signed consent from her" placed ownership of genetic information with the family member.

Other controversies related to family perception of ownership of knowledge such as granting rights of access to decisions of extended family members. An early set of

notes contained a beautifully annotated and hand drawn family tree dating back to the 1600's with an accompanying letter. In contrast with the medical pedigree noting symptoms of early cataracts and age of death the family pedigree recorded family names and social recognition or awards. The letter also made reference to "...supplying complete genealogical tree...If you could let me know without mentioning names whether any tests have been carried out on the progeny of my father's brother...". The inference that providing information on family members gave access to information about their medical status required careful response outlining the personal nature of medical information without causing offence.

DM classification is embodied in the person and its presence was apparent sometimes without their awareness. The "myopathic facies", and grip myotonia evident in a handshake could be elicited without a medical examination. Featherstone et al. (2005, p. 554) refer to the way that "Appearance has long been thought to reveal the inner character of the person". Lack of facial expression due to muscle weakness and extreme tiredness are part of the classification of DM and are associated with additional classifications such as apathy. Bowker and Star (2000, p. 26) described how a "cage formed by classification systems can be constraining". The notes show how "classificatory systems themselves can be understood to hide more than they reveal" (Latimer et al. 2006, p. 604). The intersection between classification and personal biography indicates how a classification cannot fit everybody and its power to categorise a person. Yet the notes contain traces of how actively family members, even those most affected by DM, worked at reaching an understanding of their condition.

The converse of DM classification is the disentangling of other conditions from it and the recognition that DM is not necessarily the cause for every sign or symptom. The notes document instances where the geneticist was active in raising awareness in health professionals of the need to consider other diagnostic possibilities for additional health problems

An aspect of classification lies in its relationship to the practice of contemporary medicine. The process of diagnosis and then treatment is central and reliant on an underlying definition of disease. In the case of DM this was recognised but not

clearly defined at the genesis of the archive. The field of medical genetics was itself emerging as a shift in thinking about the causes of disease. This was apparent in the earliest notes where there was one geneticist, no laboratory staff or separate department. DM was poorly recognised until it was given greater visibility by the gene discovery, although the family interviews indicate the continuing variability in knowledge outside medical genetics as a source of stress. As knowledge about the condition increased and advances were made in defining it as a single gene disorder the Institute began to expand. This was not solely due to DM, but the medical records for DM trace the development of knowledge alongside acquisition of greater recognition and resources for the Institute. Classification plays an important role in the profile and recognition of professional identity. Assertion of skill was visible in the interpretation of a clinical examination where the opinion of a referring doctor was politely refuted through correspondence.

Her grip was fair and <u>not</u> myotonic.

Physician to GP cc. Prof 1975

Myotonia of grip <u>present</u>

Prof to physician 1975

Association with specialist knowledge and expertise, in addition to academic work such as publications contributed to this expression of identity. The profile of Prof and the Institute was conveyed by this 2002 letter from a geneticist in another area to a paediatrician where he comments that "There is an excellent clinical genetic service...where [Prof] has a special interest in and unique experience of md..." Publications relating to scientific research, genetic counselling and practical management reinforced the reputation for excellence of Prof and the Institute. Contemporary notes indicate a continuation of this reputation with regard to the DM clinic but resources associated with DM scientific research lessened in the years after gene discovery and illustrate the impact of research reputation on service provision.

The way classification systems become an invisible part of an institutional infrastructure has been noted (Bowker and Star 2000). Where classification is functional it becomes invisible and difficult to analyse (Bowker and Star 2000, p. 33). Practice reinforces classification and embeds it in institutional identity. The notes document how this practice was handed down through generations of junior doctors, many of whom became members of the culture of clinical genetics. The

notes shaped this process through documenting what had gone before and providing a template for the future. The emergence of the DM protocol from the handwritten and bullet-pointed lists that preceded it show how classification arose from iterative practice until it eventually became part of bureaucratic as well as clinical identity. In this way it exerted control over what constituted a DM clinic appointment. Through classification information could be gained about the past such as how previous geneticists had carried out their work and how this directed the work of future geneticists.

The maintenance of classification through membership and iterative practice is associated with families and professionals. Becoming part of DM as an inherited condition entailed activity on the part of the family in reconfiguring their past and learning a new linguistic repertoire to express their new classification and communicate in the culture of medical genetics. Classification in the DM record is informational and organisational. It is a fundamental activity within the notes and functions "by highlighting certain properties, downplaying others, and hiding still others" (Lakoff and Johnson 1980, p. 163). This could be seen in the iterative process of identifying certain signs and symptoms such as myotonia and a "typical myopathic facies". However categories are not fixed and can change according to the context of the time. This necessitated the mediation and interpretation of information coming from other professionals

> *I was most interested to see this extraordinary family...I think this must be regarded as one of those very rare cases of mutation to this extraordinary condition which afflicts [mother] and almost certainly afflicted [deceased child]*
>
> *Referral from out of area geneticist to Prof 1978*

> *It certainly sounds as if Mrs...might be a new mutation but I think one would have to have normal slit lamp and EMG studies on the parents and apparently healthy sib before being certain of this.*
>
> *Response from Prof to out of area geneticist 1978*

The notes trace how behaviour with regard to diagnosis was shaped by context so that before gene discovery diagnosis was based around clinical examination, EMG and slit lamp examination and after gene discovery was shaped by protocols regarding genetic testing.

Hammersley and Atkinson (1995, p. 173) refer to the function of documents in establishing actors as 'cases' with situated identities, which conform to 'normal' categories or deviate from them in identifiable ways". The process of making a diagnosis and of researching DM constitutes case- making work. Before the gene was discovered the process of making a diagnosis was also linked with establishing knowledge about DM itself. The translation of clinical observations into intellectual material was accomplished through language and a subtle change of definition. Through consultation the person became a patient and a process of depersonalisation transformed them into a case. By equating the person with the condition the engagement was now with aspects of an impersonal label, rather than a patient.

The potential for clinical observation to became significant in research was not limited to the geneticist and particularly before gene isolation the notes document the referral of the "curious" or the "interesting" case.. In the early 1970's Prof established a Study of Infantile MD in Britain that took the form of a typed report and was sent to paediatricians, neurologists, GP's and other clinicians considered likely to see DM in their practice. The observation made by a paediatrician of a child who "...was one of the original cases of congenital myotonic dystrophy described by Vanier in 1960" illustrates classification in practice.

The importance of clinical judgement was evident in cases before gene discovery where results were interpreted as being equivocal. Once the gene test became available clinical examination was carried out, but the definitive diagnosis was arrived at through the application of technology rather than clinical judgement. In many cases the diagnosis was apparent but the gene test offered confirmation and certainty.

> *I have no doubt that she was clinically affected...We discussed the relationship between her symptoms and the diagnosis of md and I took a blood sample for molecular genetic confirmation of the diagnosis.*
>
> *Geneticist to out of area geneticist 2004*

However there were still cases where clinical skill seemed to suggest one possibility only for this not to be confirmed by the blood test. For the families the gene test represented certainty of their diagnostic status and the first instances of a family member requesting a gene test but rejecting clinical examination until the outcome of the test result appeared. The implications for the DM clinic lie in the potential of a

reliance on technology to undermine the decades of experience and learning that underpin clinical examination and clinical diagnosis of DM.

The punctuated chronology of DM: Intersections of everyday life and work

Introduction

A large set of notes, sampled at the end of the data collection, marked the first referral and the initial medical record in the DM archive. Their analytic significance lies in the connection between three generations of one family: Ben, a toddler, his mother Alys, and grandmother Delia, with the discovery of the DM gene and Prof's career at the Institute.

These retrospective narrative trajectories document the emerging classification of congenital DM, and offer insight how research was accomplished through cooperative relationships with families. The significance of the notes within their own culture was implied by the handwritten instructions on the front indicating that they should not be destroyed.

The use of a single set of notes was used to supply "an alternative account in which human experience and understanding, rather than objective truth, played a central role" (1980 Lakoff and Johnson, p. x). The analysis focuses on the construction of the referral as the inherited condition of DM, the use of rhetoric in research, the case study, and the relationships that were integral to research and management of DM in the family.

In taking the "unit of narrative" as a family member the analysis represented the richness and emotional intensity of the notes and the way in which they brought to life both DM and the everyday life of the family beyond their intersections with service and research. The temporal aspect of the notes allowed the way research was characterised by intermittent contact with families to be fore-grounded, and most significantly how it was people with their motivations and their relationships that characterised the way progress was made, rather than procedures or protocols.

Constructions of reality

> *I asked [Prof] to see them that afternoon as the mother [Alys] is 18 weeks pregnant and admitted that she had deceived you on that point.*
>
> *Neurologist to Prof and paediatric neurologist 1972*

The first referral to Prof, newly arrived to set up a department of medical genetics, exemplified the complexity of reproductive risk and the interpretation of professional and family dynamics that characterises the practice of clinical genetics. The referral introduced the diagnostic complications surrounding Ben, the toddler who had been introduced to Prof by a paediatrician following a talk he had given on DM. The introduction of DM as a possible diagnosis for Ben reframed the context for the family in terms of reproduction and his future. It also reframed the diagnostic question for other professionals

> *I agree that the mother [Alys], clinically, showed some features of DM, although these were mild...She had a myopathic facies with some proximal girdle weakness...I sampled only one muscle in the child [Ben], and note with interest that my examination on the last time was confined to the lower limbs...In view of these findings I did not think it worthwhile examining the father.*

The above letter from the neurologist to Prof indicated change in the application and interpretation of technology. Looked at using Labov's (1997) structural approach this referral contained the explanation of what the overall narrative is about, the people involved, the time it began, and the arena in which it would be played out. The diagnosis of DM in Alys led to Ben being re-evaluated in the light of emerging knowledge. The language, in particular the use of the word "interest" matched observations across the whole data set that it was used consistently to signify the unusual, the not-fully-understood, or the exception to the rule. Here the context of the neurologist reviewing his diagnostic procedures and the cooperation necessary to make a difficult diagnosis was highlighted. From this common beginning the analysis focuses on the narrative trajectories of research and service as it impacted on the family and on Prof.

Research involving Ben led to the classification of congenital DM and the subsequent explanation of the phenomenon of anticipation through the triplet repeat expansion at gene discovery. Analysis explored how this research was accomplished through the relationship set up between Alys and Prof. The relationship between three generations of the family and the management of DM gives insight into

changing practices and also dynamic interplay between family members who chose not to have a diagnosis despite manifestations of DM.

The brief introduction of the father and husband as being irrelevant to the diagnosis and classification was symbolic of his presence in the notes where he remained invisible other than through unresolved legal letters regarding a possible divorce. He reappeared in the handwritten notes documenting the death of his son Ben thirty years later.

The construction of four family trees at the front of the notes and an additional small pedigree filed later indicated the context dependent nature of interpretation. The small pedigree names Sean, the baby who died following the second pregnancy, and notes an abortion that has no reference elsewhere in the notes. The presence of another inherited muscle disease in a different branch of the same family was the subject of two family trees marking the two conditions in different ink. The addition of extra details onto the family trees indicated the incremental nature of the knowledge being collected. An undated three-generation study was also filed and the notes convey a continual movement between the present and the past as new information was gathered and interpreted.

Rhetoric in research

> *A recent development with this disease is that it is now possible to make a pre-natal prediction by amniocentesis based on the secretor type of the parents and the foetus.*
>
> *Prof to paediatrician 1972*

The connection between research and the narrative of the family was apparent from the first entry. The referral of Alys for genetic counselling for her second pregnancy, and the realisation by a paediatrician that Ben, her eldest son, may have DM linked research and service. Wright Mills (1940, p. 904) noted, "The differing reasons men give for their actions are not themselves without reasons". The way families were approached for research was visible in the notes through letters. These letters were essential for recruitment, and analysis involved examination of the accounting devices they utilised. The first of these letters appeared three years after Alys had been diagnosed and asked "for your help in the work I [Prof] am doing on your particular condition." The personal request linking the significance of DM for the

family with research was followed with a question as to whether a home visit was possible but concluded with "If it is not convenient please do not hesitate to let me know." The initiative required by Alys to opt out rather than make contact to take part inferred a mutual interest in research.

The main research focus was on Ben, and research access to him also invoked a cooperative and positive construction of research. It demonstrated "the ways in which linguistic elements are organised to persuasive effect" (Fairclough 1995, p. viii) and how language was central to the maintenance of the research relationship between families and professionals. The justification for contact was based on the shared understanding of the importance of research. Vocabularies of motive, such as duty to others to research an incurable hereditary condition, provided connections between families and clinicians, and demonstrated how "When they appeal to others involved in one's act, motives are strategies of action" (Wright Mills 1940, p. 913). The persuasive power of discourse lies in its ability to change behaviour but power also resides with those who can decide whether to participate or be persuaded (Lakoff 1982). Although Alys cooperated with research throughout her involvement with the Institute there were many other DM medical records documenting repeated unsuccessful attempts to recruit families. The notes also illustrated the link between genre and text, with handwritten bullet point lists recording clinical examinations and summaries of test results for GP's contrasting with the engaged tone of the research request or the explanatory focus of the DM clinic letter to the family.

The narrative of clinical and scientific research leading to gene discovery was characterised by periods of activity and contact followed by periods of quiescence.

> *I have been asked to write an article for a medical journal on [Ben's] disorder and thought it would be very helpful if you would allow me to include one of the photographs of [Ben] and yourself ...This would be of great help to doctors trying to recognise this condition in other patients and it would only be doctors, not lay people, who would read the article...*
>
> *Prof to Alys 1976*

The reference to the article being accessible only to a professional audience was comparable with similar observations in Lindee's (2005) study of research into the Amish culture. Lindee ascribed it to the cultural norms of the time where it was assumed that professional interests operated in a different domain to those of lay or

patient groups. The way impression management was accomplished linguistically could be seen in the reference to expertise in writing the journal article. References to expertise performed identity work in professional practice and could be seen in referrals to Prof as "the local expert" shortly after his arrival, and also in later referrals by the DM geneticist seeking "expert opinion" from another specialist.

Requests for assistance with research continued but there was also evidence of a service commitment and of attending muscle clinic. The narrative of disease progression was recorded primarily through this contact. The discovery of the DM gene resulted in the availability of a gene test, although the status of Alys and Ben had been confirmed clinically beyond doubt. Extensive knowledge of DM through relationships set up with families to document its natural history formed a vital part of the contribution to gene discovery. The narrative of hope and the significance of clinical understanding of the condition extended to key professionals such as the family GP as justification for monitoring its progression.

> *Recent advances in understanding this disease means that there is the possibility of specific therapeutic agents in the not-too-distant future and we are trying to document the untreated course of the condition systematically so that, when any treatment does come we will have a proper baseline for it to be introduced.*
>
> *Geneticist to GP 1993*

Ethics and the development of protocol arising from clinical practice were also evident, such as written consent for the photographs filed in the medical record. The emergence of ethical codes of practice was visible in a later entry regarding a research request for a test result.

> *As that is one of the incidences for which we have not yet devised a consent form, I wonder if you would mind sending him a note to give your consent for this (if you still agree to it)*
>
> *Junior doctor to Delia 1994*

Once the gene had been discovered the research requests changed in nature and varied over the next fifteen years from research into hearing to studies on sleep and heart disease. The series of junior doctors were no longer apparent after 1996 and research projects were primarily introduced by the geneticist at the specialist DM clinic rather than instigated there, and indicated a move away from molecular investigations.

We are carrying out a study and looking at levels of sleepiness and possible associated difficulties experienced by people who have md...

Clinical psychologist to Alys. Undated post-gene discovery

...information about a new study in md and heart disease...contact if prepared to take part...

Geneticist to Alys 1999

The notes were divided into three sections, one for each generation, although there were occasions of overlapping and duplicated information reflecting the familial implications of clinical genetics in practice. Towards the end of the section for Alys there were traces of involvement with the DM research register. This illustrated how the framework for accessing family members for research purposes had been formalised in the years since Alys first cooperated with Prof. The relationship between Alys and the Institute continued until her death despite changes in research focus and protocol and highlighted the way the tradition of research was integrated, alongside service provision, into the developing Institute from the first referral

The case of a life and the life of a case

...Dr G showed myotonic discharges on EMG on both her [Alys] and [Ben]. In addition [Ben] showed the characteristic facial diplegia of the infantile form of the disease.

Prof to paediatrician 1972

The diagnosis of classical DM in Alys and the simultaneous diagnosis of the childhood form in Ben, her first- born son, was the clinical manifestation of the phenomenon of anticipation. The significance of the first referral for DM lay in its link with research, although the immediate focus was in genetic counselling for the young couple. The birth and almost immediate death of Sean, their second child, due to DM, was noted in the medical record. The DM archive documented how Prof embarked on a national study of congenital DM and the research focus on Ben was highlighted in approaches made to Alys. The way the little boy became an exemplar of a new classification was traced in the notes through his representation as a case. The work of categorisation, defining and sorting through language reflected the intended audience.

...I was struck by the improvement in his general development since I last saw him a year ago...He has considerable speech though it is still very indistinct, is active and co-operative, but still turns his left foot inwards...

Prof to paediatrician 1976

At this point Ben was being managed by the paediatrician, but the focus of the next entry in the medical record emphasised the academic interest of the case. DM as represented in the clinical setting by Ben was now being described molecularly for educational purposes and in this process the language became less personal.

> *A two year old boy was seen because of delayed motor development and unusual facial appearance...There is usually profound facial and jaw weakness, giving an expressionless "carp-mouthed" appearance.*
>
> *Prof case presentation 1976*

In the first letter to the paediatrician Ben was presented by name. The language implied action, indicating what Ben could and could not do, and the presentation was that of the impact of the signs and symptoms of DM on him. In the later entry the letter documented a case for academic and research purposes.

The language was that of classification and again "interestingly" was used to highlight an observation of potential significance. The representation of Ben as a passive exemplar was further highlighted by the way in which he was offered as a case history to another colleague in the form of a photograph.

> *This is the boy about whom I spoke to you before...a classical case of myotonic dystrophy, presenting at birth with hypotonia and facial diplegia and interestingly also having a large head...I hope the muscle samples prove suitable for histochemistry...It would be very nice to be able to include suitable photomicrographs as illustrations for my book as I have used him as an illustrative case history for the congenital form. Please feel free to include him in any series for your own purposes...*
>
> *Prof to geneticist in out-of-area specialist muscle clinic 1978*

Anspach (1988) drew attention to four key features in work on case presentations in the clinical setting which were: de-personalisation, use of the passive voice, treating medical technology as the agent and placing emphasis on the subjectivity of the patient account. Although they relate primarily to presenting to an audience these are of interest in contrasting the two accounts. In the case presentation account Ben is presented as the passive representation of DM. It was the disease that commanded attention rather than the person with the disease. In discussion of the muscle biopsy Ben is further transformed by a senior technician into "some nice frozen sections" that "stained well" and "should photograph nicely". This de-personalisation related to the academic and presentation aspects of DM and of Prof, as the book referred to in the excerpt was part of his own process of building a career. Ben's photograph

gave a visual representation of his status as exemplar of the congenital DM classification, and insight into the professional maintenance of relationships.

The contextual nature of narrative and the way it involves sense-making and "has close links with narrative, since it involves retrospective accounts that stress plausibility, credibility and coherence" (Fairclough 1995 p. viii) was visible in the varying reconstructions of the little boy. The presentation of his function in relation to everyday life contrasted with the case presentation style in choice of language and agency. The contrasting of DM classification in theory with classification in clinical practice was embodied in Ben as he grew up and his representation as an exemplary case was implicit in correspondence.

The narrative of a person's survival in the face of unfavourable circumstances was clear. DM was placed as an obstacle within the broader context of a life and the language was of surprise and a suspension of disbelief that normality could be achieved.

> *His main limitation centres around his myotonic dystrophy with profound weakness, but in spite of this he is able to lead a surprisingly normal life...*
> *Geneticist to cardiologist 1997*

> *In spite of his handicaps he, apparently, goes swimming twice a week...*
> *Psychologist to junior doctor 1988*

The referential nature of the trajectory of Ben's life with the trajectory of DM was conveyed through phrases such as "remarkably well" and "considering the severity of his problems."

Prof instigated the practice of writing directly to the family, in addition to the GP, after genetic consultation at the start of the clinical genetics service. Through the notes it became a standard part of practice that subsequent generations of geneticists followed. The importance of audience and context for documents could be seen occasionally through contrasting accounts of the same clinic in the family and GP letters.

> *This is just to go over the things that we discussed when you came to muscle clinic recently. I am sorry if all the questions that we asked upset you.*
> *Geneticist to Ben 2000*

This pt [Ben] was seen in the muscle clinic recently. He was not very co-operative.

Geneticist to Ben's GP 2000

The significance of the little boy to research, and to Prof, continued throughout his childhood and adulthood.

This man [Ben] with congenital myotonic dystrophy and associated mental handicap has remained essentially well over the past year. It is interesting to note that he is now...years old and that he was one of the first cases in the UK to be diagnosed, having been seen by me at the age of 2 and followed ever since...

Prof to GP 1998

The entry recording Ben's death as a result of falls referred to his early diagnosis and management by Prof over his lifetime.

This 32-year-old man was known to suffer from CMD. He had been a pt of [Prof] in [the Institute] for many years, having been diagnosed with this as a young child, in fact 2 years of age...

Autopsy report 2002

Research was the dominant theme once again as consent was sought from Alys for tissue donation. The handwritten entries by the DM geneticist while Ben was in the intensive care unit provided insight into the complexity of this decision for Alys and her continuing commitment to research.

...agreed in principle to the collection of any tissues yesterday. On reflection prefers no tissue samples from above the neck...Reassure that she is able to specify exactly what she is happy to allow...She was clearly very distressed and bereaved but still quite clear she wishes to help with research...

Geneticist handwritten entry 2002

The central importance of classification in the contribution of clinical expertise to gene discovery was embodied in the medical record from Ben's referral as a toddler until his death thirty years later. The notes give insight into how this classification work was accomplished and how exemplifying a classification was fore-grounded throughout Ben's life.

Life stories

It is the first time I have seen her since the recent death of [Ben] who as you know had severe cmd but survived to the age of 32. I have now been seeing [Alys] for over thirty years and it has been sad to see her change from essentially asymptomatic to now being severely affected with the condition.

Prof to GP 2003

The narrating of the course of DM, in addition to the documenting of the key relationships between the family and Prof, was integral to the construction of the above letter. DM was the central focus around which the notes were constructed. It was mediated through relationships, measured through procedures and protocols and narrated through handwritten notes, letters and laboratory reports. It was implicit in the management of the relationships between the DM clinic and Delia, Ben's grandmother, who resisted diagnosis, and was central to the management of Ben and Alys who symbolised the foundation of DM research and service.

The span of the notes gave insight into the multiple representations of DM through correspondence from agencies as disparate as physiotherapy and day centres, to cardiac management and allowances. The impact of DM itself as recorded through annual measurements was contrasted against the recording of Alys declining "having any particular problems at present". This contrast was apparent in the family interviews and suggests that it was agency and ability to carry out valued tasks that defined the personal impact of DM.

> *For many years her [Alys] life revolved around caring for [Ben] and now that she is on her own, I think that she is very isolated and probably clinically depressed...my reading of the situation is that [Alys]...is probably now unable to cope with independent life.*
>
> *Prof to GP 2003*

The physical and emotional deterioration in Alys's situation was recognised following Ben's death, as was her role in caring for him. The way Prof framed this deterioration in a narrative of Aly's life emphasizes the way in which DM in the medical records was represented in terms of its effects on everyday life. It illustrates how it is not only "patients " who tell stories and that "the purposeful reconstruction of past events across time, is an essential vehicle for talk about illness as much within the medical institution as in the 'lay' community" (Gwyn 2002, p. 141). Murray (2008, p. 14) defines the main function of narrative as being "to bring order to disorder. In telling the story, the narrator is trying to organize the disorganized and to give it meaning". Ricoeur (1987, cited in Smith 2008, p. 114) described the ongoing tension of this attempt to make sense of everyday events and of how "concord cannot be without discord". Alys's refusal to contemplate formal care illustrated the complexity of decision-making and the central importance of agency

that she had asserted at key times such as her initial decision to continue with the pregnancy following the first referral.

The central importance of family dynamics to the research and service of DM was evident in the incorporation of the needs of Delia, the grandmother, despite her refusal to be diagnosed, into the management of her daughter and grandson. The tacit acceptance of DM, implied through her attendance at clinic and donation of blood, was accepted also by the DM clinicians until she became seriously ill.

The establishment of classical DM in Alys and CMD in Ben were achieved through clinical diagnosis and confirmed molecularly after isolation of the DM gene. The progression of both forms of DM was documented and gene discovery did not alter the diagnosis, only the nature of subsequent research projects. However the significance of the gene test lay in the resolution of "the clinical suspicions" about Delia, where "evidence" over thirty years had been inconclusive. Her mild symptoms had led to "an assumption in our family notes...she [Delia] was very mildly affected...The family however have always denied that she [Delia] had this problem." Delia's decision to have a confirmatory DM blood test was narrated as part of concern about appropriate medical management but also resulted in the molecular confirmation of the three generations of the family as representative of the phenomenon of anticipation.

The intertwining of Alys's life, the career of DM and Prof's career was integral to the notes. There was a small reference made by another geneticist on a DM clinic record sheet that both Alys and Ben preferred to see Prof. This was one of the few occasions where they made their wishes explicit.

Temporal framing served to emphasise the significance of this particular medical record. The trajectory of the career of the geneticist was directly linked to the lives of Alys and her son Ben. The potential significance of this may be intuited from the GP letter in which Prof alerts the GP to the fact that he will no longer be seeing Alys and Ben.

I have now been seeing [Alys] along with [Ben] for over thirty years...I am arranging to see her again in the early summer, which will be the final time she sees me before I retire.

Prof to GP 2003

The only handwritten letter in the notes was from a relative informing the Institute of Alys's death and referring to her involvement with DM research.

Dear Sir, ...My cousin [Alys] passed away...at ward...From some of her letters we note that she suffered from MYOTONIC DYSTROPHY and could have been involved in a study of this complaint...

The final letter in this narrative was from the DM clinic geneticist in 2005 to Alys's cousin, replying, "I am very sorry to hear of your cousin's [Alys] recent death. I would like to send the condolences of the team to you and your family..." and including details of the MDSG.

The narrative, which began with the young Alys pregnant and realising there was a history of DM in her family ended with her death. Prof who had seen her originally had retired and from a single person establishing the original department the condolences to her family were now expressed by a team.

Summary

Analysis of the DM archive traced the active work of documents in the way "They are constructed in accordance with rules, they express a structure, they are nestled within a specific discourse, and their presence in the world depends on collective, organized action" (Prior 2003, p. 12). Through exploring relationships, risk and classification the many representations and interpretations of DM were explored. The linguistic devices used to communicate DM as a hereditary and incurable disease, and this relationship to both service and research were explored through analysis of the use of metaphor. Insight into how the processes of service and research were underpinned and mediated through relationships was explored in a single case study. Central to the analysis was an exploration of the characters behind the processes and procedures that define institutional identity.

Contemporary records show how the referral process has become bureaucratised with protocols indicating management and diagnostic pathways. However once the process has begun it involves family history and the construction of a family tree. On

confirmation of diagnosis, management options are then discussed and the possibility of attending the specialist DM clinic introduced. The contemporary protocol shows how even where an individual has been referred and had a diagnostic test the implications of DM continue to be reflected in the wider family, and the skill and expertise needed to negotiate family dynamics remain a fundamental part of the process. A theme across the notes pre and post gene discovery is the direct communication through letter with families following consultation and the framing of issues according to the family point of view. The analysis highlights how diagnostic uncertainty has been resolved since DM gene discovery, but that uncertainty has moved to focus on prognosis.

The notes demonstrate that categories and classifications have to be activated as well as constituted. The black box metaphor has been applied to the construction of medical disposals in clinical practice Berg (1992, p. 154). The way questions are constructed and selected influences the outcome and this applies equally to the questions that are not asked. Prior to gene discovery the classification of DM through the building up of clinical expertise was based on the constant iteration of "typical" signs and symptoms" leading to a characteristic presentation. The DM medical records document many home visits carried out by Prof and the junior doctors prior to gene discovery. The current DM protocol developed from the format of these consultations. In the contemporary DM notes there are also records of home visits carried out by genetic nurse specialists specifically for genetic counselling. A different profile of DM emerges, elicited by the specific purpose of the visit. These representations are equally valid but indicate how different discourses are shaped by the context in which they occur. The profile of the muscle clinic has declined in the years after the definitive phase of gene discovery and although the DM clinic is recognised as a centre of expertise it is also part of an NHS service where resource allocation is competitive.

The pedagogical function of the notes was implied through the series of junior doctors who documented research and management of DM. Pettinari (1988, p. 131) likens this to a "folk practice" whereby "This type of reporting is rarely formally taught, nor are the rhetorical acts of describing findings and describing procedures commonly formally presented". Format and classification were fundamental to this

activity, with specific observations and measurements performing core functions in making DM visible as an inherited neuromuscular condition.

The relationship between talk and text in the written notes and letters of the DM record was captured in the introductory excerpt where the geneticist referred to the consultation with the family member as "your story" and repeated information in order to accomplish the transformation into DM. It indicates the potential of story telling as "the preferred sense-making currency in organisations" (Boje 1991, p. 106). The rhetoric of the consultation became transformed into a written activity, in addition to DM becoming transformed into a genetic narrative.

Flower (1981, cited in Pettinari1988, p. 89) distinguishes between texts that are writer-based and "organized according to a logic based on the writer's own needs", and reader-based, "focused around information that readers want to obtain". The clinic consultation letter is structured around conveying information to the family and this is accomplished linguistically by references to "you" and "your story". It shares some features with therapeutic listening in a counselling activity, where details are reflected back to indicate that attention has been given to the speaker's account. In the written format this also conveys a new and mutually constructed reconfiguration of the clinical history into DM.

The medical records, particularly in the years leading up to gene discovery also documented research related correspondence that was interpreted as "writer-based" in the initiation of relationships governed by a specific interest on the part of the geneticist. These letters were characterised by references to Prof by the junior doctors as a way of establishing credibility, and in the earlier letters by Prof's references to books or articles he had published. This became less necessary as the departmental profile became established and the letter itself accomplished the function of conveying seniority and status through titles and letter headings. The motivation of individuals that lay behind the accomplishment of both service and research development can be seen in these letters that are driven by personal interest rather than imposed by external management structures.

The notes document DM as a classification but within the correspondence from geneticist to family there is variation in narrative styles. This ranges from extreme detail conveying possible scenarios and communicating the characteristic uncertainty of DM, to letters summarising the outcome of the clinic consultation. This difference in detail is particularly notable in the letters from the DM clinic geneticist who also completes the DM protocol, and geneticists seeing families in regional clinics who uniformly address aspects of inheritance but vary in their focus on manifestations of DM in everyday life. The notes capture these various discourses and illustrate the central importance of context.

The episodic nature of recording has been reflected in the analysis of the punctuated chronology of DM (Pettinari 1988). The notes are a partial representation of DM and do not claim to present an exclusive picture. They present DM as a trajectory where the management of uncertainty is formalised and where everyday life intersects with the documenting of a degenerative condition, such as times of reproductive decision-making.

The documenting of DM itself over time in the archive is an ongoing trajectory, with every consultation, test and report leaving a trace that emphasises how much has been learnt about DM but also those aspects which remain poorly understood, despite gene discovery. Within the archive there are multiple trajectories from professional and family perspectives, of what it is like to work with and to live with DM. The DM records retain the past but also the present in the documenting of the clinical consultation, where it is the relationship between the geneticist and the family member, with all the implications for extended family and uncertainty of the prognosis of DM, which remains unchanged as the core activity.

Chapter Six

Gripping Stories: Narratives of DM and everyday life

Introduction

Stories of encounters with professionals, and of family and personal relationships characterised the DM family interviews. One young man, Alan, speaking of how he was asked to tell medical students about DM, highlighted this.

> *I said "I don't know how to explain it in words but I can show you" and the way I showed him...when I couldn't let go of their hand they realised what I was talking about, about muscle stiffness.*

Grip has both a physical and metaphorical symbolism in DM. Grip myotonia is a key diagnostic feature of DM, manifested in a difficulty releasing the hand. Alan's demonstration for the medical students served as a conduit into the DM domain though another metaphor of communication, that of the handshake.

> *I can't grip anything, many things these days but if I do grip somebody's hand a bit too hard my hand won't relax and I just can't let go. So when I, I mean, when you shake hands with a man, men grip. They always do, they always grip, even if they shake hands with a woman, they grip the hand. I don't grip. I just put my hand out and let them grip my hand.*

The performance of the grip manifestation, rather than its description, accomplished the intention of communicating understanding and holding the interest of the students.

The physical implications of DM meant that in many cases the interview was a challenging situation. Difficulty enunciating words and physical fatigue influenced the telling of narratives. However, subsequent analysis revealed the emotional and powerful use of language. Just as Alan communicated the understanding of grip through demonstration, its metaphoric counterpart of dealing with change was communicated through the accomplishment of narratives.

Transformative narratives communicated the experiences of being diagnosed with and living with DM. The narrative and analytic themes were found in the broad categories of DM, relationships and the self. The analysis explored how these themes were represented through narrative accounts of change. The poet Seamus Heaney (cited in Tobin 1999, p. 83) referred to the way people use stories as "as posts to fence out a personal landscape". This analysis explores how DM was represented in

narratives constructed in semi-structured interviews with twenty people diagnosed with DM. The analysis recognises the richness of the data and the many interpretive possibilities open to the researcher.

These transformative narratives were rooted in everyday life and constructed a network of relationships including family and the DM clinic. The information sheet, which was included in the information pack sent to families, referred to "The impact of gene discovery on patients, doctors and their clinics." One potential transformative narrative implicit in the research question, that of the diagnosis of DM transforming a person into a patient, was resisted and the medical setting was narrated as one amongst many arenas where identity was represented. Transformative narratives refer instead to the process by which DM was integrated by family members into a reconfigured sense of self through attribution of personal meaning. This was narrated through accounts of the challenges to valued roles and identity in everyday life and the awareness that if change itself could not be predicted then attitude to change could be.

Repertoires of narrative genres, interpretive registers and linguistic choices reflected personal choice and access to the narrative resources available. Analysis highlighted the prevalence of highly emotional language. Thematic analysis allowed exploration of where this language was concentrated. All of the categories were acknowledged as co-constructed as reference was made in broad terms by the researcher to how the person became aware of DM, family relationships, and the meaning of DM on a personal basis. However themes arose out of iterative and reflexive practice where the experience of initial interviews informed subsequent encounters and issues of apparent significance were repeated throughout the interviews.

Family members used vivid and immediate language to narrate their experience of DM, utilising metaphor and emotion to convey turning points in personal biography such as diagnosis. Contrastive rhetoric functioned to convey the intersecting trajectories of everyday life and the progression of the condition. DM was contextualised in relation to past experience and future expectations. The family members constructed identities that emphasised roles of everyday life and narrated past accomplishments that were no longer possible. Uncertainty about the future

related to the manifestation of DM and uncertainty about the loss of anticipated roles and stages such as active retirement, parenthood and employment. Moral tales conveyed good and bad experiences in work, the clinic and personal relationships. Agency and personal meaning moved the accounts from situations grounded in everyday life to the process of constructing a way of dealing with the uncertainty of the future. These varied in the extent to which this had been accomplished but the narratives were distinguished by awareness of a need to establish personal meaning.

There were particular accounts which were found in all of the interviews relating to perception of DM itself, finding out about DM, negotiating everyday life with DM, reconfiguring the family and comparisons with both the past and the imagined future. Accounts related to the search for knowledge initially and later for meaning.

"Oh God, here we go": Accounts of Diagnosis

The mapping of the trajectory of DM alongside a life trajectory featured in stories of diagnosis. In some interviews people returned repeatedly to the event of diagnosis itself, using phrases that had been said at the time and structuring the diagnosis as a defining episode. Even where the diagnosis was made in childhood the diagnosis narrative communicated a sense of being defined by DM, a theme that was returned to over the course of the interview.

> *For as long as I can remember I've known about it, because I was only a few months old when dad was diagnosed, so I've always known about it and I mean I wasn't diagnosed until I was nine years old, but I've been going to the hospital every six months for as long as I can remember...I used to go with my father, he used to go and see Prof and I used to go and see [junior doctor] and (indiscernible) and, so you know, I knew. I didn't exactly know why I was going because there was nothing wrong with me then but they wanted to keep an eye on me basically because I was male. I know obviously women do get it but women are more likely to be carriers and men can't carry the gene they have the gene.*
>
> *Alan aged 33*

Alan illustrated how DM could define a family through contrasting the impossibility of finding a role within his family, with the happiness of being accepted as a visitor in another family who "forget I have DM." The significance of being male and having DM was recurrent in his later narratives of searching for a defining role of being a man within the family.

The diagnosis story was reflected upon from different perspectives throughout the interviews. It had a reverberative effect whereby the description of the event itself, often recalled with exceptional clarity, was then used as a key focal point in relation to other significant issues.

> *When I was twenty six*
> *What they actually found out was my mother had cataracts.*
> *They're a certain type of cataracts which everyone in my ancestral history has these cataracts.*
> *And they asked if anyone in the family had any problems with mobility and things like that.*
> *And she told them it can trigger in a lot of cases (inaudible)*
> *And they said. Oh, send her up.*
> *So they checked all different things, asked different questions. And said that 'Yes', so I occasionally go to the eye clinic to have those checked down there.*
> *Sort of MOT every year.*
> *But that's how they actually picked it up.*
> *And I was twenty six then.*
>
> *Maria aged 46*

The structuring of the account followed Labov's (1997) classic format. Of interest was not simply that family members chose to present their recollections of diagnosis in this form, but what they included in their accounts. The temporal aspect of giving an age, or a year, indicated a seminal or turning point and in the interviews acted as a signifier for the events and changes that followed. The language used was emotional and vivid, with images of shock, loss and reinterpretation of self and family. The following is a composite of the language analysed in relation to diagnosis, which reflected the differing backgrounds and routes to diagnosis but shared an emotional intensity.

> *It all came out of the blue... shock...pretty upset actually...I went back to work and broke my heart...So I didn't handle it too well...it hit me...devastated...It was the last thing I expected...it's a real shock...just surprise, you know... ...I just sobbed my heart out...It was the worst minute of my life so far that was...well, I was devastated, absolutely devastated, I thought no, this - I haven't got this thing but -...I was just in a heap on the floor, like it was horrendous...Oh god, I got this thing...I cried for days...And he said I - you were a bit shocked to find out. Shocked was an under statement. I was devastated... I was reading a book that I bought with that you know Prof. But I mean nothing was sinking in. You were just re-reading it and re-reading it. You just (inaudible) really you were trying to bury your head...It was just when that doctor called me in and I thought well there's something...Oh God, here we go... I knew something then...Oh, there was definitely something wrong, I always knew there was something wrong, I suspected...It's so unfair though.*

Family relationships, personal symptoms, and the accomplishment of everyday roles were evaluated and re-evaluated in the light of this common account of diagnosis. Leo, now sixty one, recalled how "... I was aware during my teens that I had difficulty getting up in the morning and it was only when I reflected on this I realised that I had had it from the beginning really..."

The seeking of new points of reference in the shape of knowledge, and stories of the transformation of old reference points such as relationships and previous interpretation of symptoms in the light of DM, illustrated the mapping of DM onto existing identity. The interview narratives communicated how the diagnosis of a genetic condition was played out in practice. Diagnosis and classification of DM led to new perspectives on individuals and their family members. Leo remembered how "I was absolutely devastated and it was the same week that I found out from my mother that she had been adopted and I had never been told."

There were positive and negative accounts of how social relationships within the family were transformed by the identification of a shared inherited trait. This was illustrated starkly in a family where relationships were already fragmented and the linking together of a common diagnosis served to reinforce the fragility of social bonds and the subsequent feelings of isolation. In this instance diagnosis did not connect the relationships in a positive way and reconfigured contemporary relationships in addition to valued relationships in the past.

> *If I'd had a sledgehammer I would have sledge hammered his stone on his grave. I hated my father. I still do. I – I was the daddy's girl all the way along until I found I'd had this...And if he wasn't already dead I would have killed him. I blame him now. For everything. Which I know it's not his fault, he probably didn't know, and we can't go back because he's adopted.*
>
> *Louisa aged 39*

For other families the genetic link reinforced relationships and catalysed a common familial way of dealing with life events, where "I think we were always close but we have a common bond now."

This highlighted the way that relationships are meaningful and negotiated in a social and emotional, rather than a biological way. The mediation of relationships was one of the implications of diagnosis, as other family members were potentially affected.

In some cases it became apparent that social relationships continued despite a rejection by one party of the familial diagnosis.

Diagnosis was a transformative narrative, involving physical, social and psychological reconfiguration. Its significance as a turning point was also evidenced by later accounts of how family members structured their biographies to narrate processes or stages of coming to terms with DM, or alternative accounts of a necessity to deal with the diagnosis in some way that was personally meaningful. This was not necessarily accomplished but the personal meaning of DM was narrated in all of the interviews, either directly or obliquely through accounts highlighting it. There were descriptions of the ongoing scrutiny of other family members, with advice on when to contact the geneticist for confirmation of suspicions.

> *"I think you've got myotonia". He [brother] said, "What's that?" So I said (inaudible) "You'll have to go to your doctor."*
>
> *Edna aged 61*

The narrating of childhood memories highlighted a turning point in the understanding of self and of others. This extended to the future, such as a desire to live differently with the condition, or anticipating the diagnosis of other family members who were being seen under the DM classification.

> *My dad would sit down and that was it. He wouldn't walk anywhere because he, because he just didn't want to I don't think. But I won't quit. I won't do that... I will not just sit down all day and go like a vegetable, no thanks. I don't want to know anything about that...I'll keep on going for as long as I can.*
>
> *Michael aged 50*

> *I mean, as I said, we didn't know what it was. He [dad] was like a vegetable after a while. He just stayed in the house and never went anywhere, like. He was really bad he was, like. If I end up like that - I wouldn't like it to end up like that at all.*
>
> *Gareth aged 58*

Accounts of monitoring other family members demonstrated how this reconfiguration was not static but became incorporated into the repertoire of family life. Louisa's anxiety for her son recurred throughout the interview as she spoke of how "he's not showing any symptoms is he at all at the moment. He's not. They said...he would probably show symptoms by now...We're always – we're always keeping an eye out..."

The gene test was associated with language of shock and distress, with the exceptions of Alan, who had been diagnosed in childhood, and Michael, who had been diagnosed clinically several decades before having the blood test. However Michael's account mirrored others in the way that he narrated the gene test result as definitive and a turning point.

> *They've been telling me for years...But they didn't know I had it until the last time I went...So they've been treating me for Muscular Dystrophy for years...But they didn't know I had it - they didn't - how can I explain? It wasn't in their notes that I had it, because they hadn't taken a blood test...So for what - I'm 50 so for 34 years, 35 years they've been treating me for it, so anyway.*

Even where diagnosis was recognised as a possibility through the diagnosis of other family members it was not fully accepted until the confirmatory blood test results came back. There were vivid descriptions of clinical observation and examination as a way of setting the scene.

Elizabeth's newborn child was diagnosed with congenital DM and died early in infancy. This led to Elizabeth being tested for DM but she remembered her conversation in hospital as she stayed by her ill baby's bedside.

> *She [paediatrician] come in and she said, "Oh, you've got it. I can tell by looking at you' she said. 'Your eyes are (inaudible). Your face is (inaudible). I know straight away." I said, "Well, I'm due a test." "All right, the test may be positive" she said "cos I can see by the way you look."*
>
> *Elizabeth aged 30*

Diagnosis accounts were structured in a way that moved between time frames of suspicion, suspense and confirmation. The emotional effects of this process were highlighted in the way that specific phrases or "scenes" were recalled with clarity. The narrative then moved to an emotional evaluation of the effects of diagnosis and the need to deal with the consequences.

Intensity was conveyed in phrases such as "overwhelmed". The narrative of diagnosis was returned to in many interviews with accounts of how it had functioned as a turning point, characterised by shock and in some case denial which was then actively worked through until a gradual process, infused with effort, led to coping strategies

An agent of change: Defining DM

> *I found that his disability is an inconvenience rather than an illness, because he is not, touch wood, he is never ill, you know. He doesn't get colds. I mean health-wise, it's an inconvenience that he has got, not an illness*
>
> *Marion aged 60, mother with DM, talking about her son Steve aged 28*

DM was not referred to as a disease by family members, but as a condition. Kleinman (1988, pp. 3-4) refers to disease as "...the problem from the practitioner's perspective. In the narrow biological terms of the biomedical model, this means that disease was reconfigured only as an alteration in biological structure or functioning".

The family members narrated ongoing difficulty classifying what symptoms DM caused. This lack of knowledge led to the expression of a lack of agency in people who were most severely affected.

> *My mouth, my eyes, all my insides, the intestines and everything else. It's all gone to pot. I feel like... my sister calls me a cripple. So you know if everything is going wrong, there's nothing I can do about it. I can't do anything about it crystals in my eyes, I can't do anything about the mouth ulcers, I can't do anything about my insides, I can't do anything about it. I think I'm worse than anybody who has ever had it.*
>
> *Michelle aged 37*

Most of the interviews included discussion about the family members affected by DM and those who were not. When asked about whether DM status affected relationships some of the unaffected relatives were described as lucky but there were no negative feelings expressed towards them, although the use of the word "clear" has implications of having escaped or not been found guilty, inferring that there was some shame attached to the diagnosis of DM.

Frustration was expressed at the way other symptoms were attributed to DM once the diagnosis was made. Louisa described how "we go to the doctors now and he seems to put everything down to myotonic dystrophy do you know what I mean?...Got a blister on your bottom, oh myotonic dystrophy." Again this highlighted the way in which the variability and uncertainty of the condition prevented individuals being in control of managing it.

DM was narrated as a dynamic entity and juxtaposed with illness, showing through contrastive rhetoric that family members did not wish to be defined by the role or

identity of the patient, although this was appropriate within certain boundaried scenarios such as the clinic. Beyond the clinic the need to enact the role of the patient in order to access the necessary resources to maintain valued everyday activities was narrated with ambivalence. The role of the geneticist as expert advocate was valued and contrasted with situations where family members described the contradiction of trying to remain positive about the future whilst portraying worst case scenarios in order to receive help. Mark, whose wife Rebecca has DM, expressed the physical and emotional repercussions, when he spoke of "The bed lifting there and the oxygen and you know, you have to tell so many people...And you're not being positive all the time which you should be. That is a sad fact on the thing...You know, if you could get the help..."

Interview narratives were constructed around roles of everyday life such as parenthood, employment, and relationships. Touching on each of these was a rejection of the identity of the sick person. The concept of disability was more complex and contested. Disability as a functional impairment was evoked by accounts of seeking specific assistance in specific situations, such a particular chair in work, and this contrasted with being seen as a disabled person. Michelle contrasted the ambivalence of her own situation with DM with that of her 10 year old daughter Stella, describing "I just feel that, I don't see Stella as disabled. She's got weakness in her hands, yeah, but her legs are like iron. You know I don't see her as having a disability at all, she's just a kid. But me, I have everything wrong with me". The wheelchair represented disability and a turning point in identity. In many of the interviews it was given as a point of reference for change. Information on when the progression of DM was likely to necessitate a wheelchair was commonly sought from medical professionals or from other sources of information such as the Internet or MDSG.

The need to narrate multiple versions of the self related also to frustration felt by being perceived solely in terms of DM by others. Several accounts were constructed to show how acceptance "for who I am" was valued while the acknowledgement of DM in terms of offering necessary assistance whilst not treating the person as a patient was given the status of an extremely significant and rare event.

Just to find out that I had got something that's going to go on and on and on.
Maria aged 46

Metaphor was a key linguistic device used to convey personal understanding of DM, which was described as having an independent trajectory, a velocity and motion that had begun mysteriously but could end at some unknown time in the future.
Leo's wife Teresa described how "it [DM] can go for generations and then suddenly two things sort of collide and it surfaces?" while Michael explained that "The genes gotta run out somewhere in the family". The sense of motion but also latency evoked by Sophie's comment that "Apparently it doesn't skip generations" reinforced the earlier observations of DM being described in terms of visibility but also unpredictability, where "It does weird things".

Words such as "trigger" evoked an agent of change. There was a sense of dynamism associated with DM that was apparent in observations made about other family members, such as Louisa speaking of her sister, where "from year to year it's devastating to see the difference in her."

Visibility was a key descriptor relating to the physical manifestation of the condition, and the interpretation of DM by others. Harry conveyed the challenge of communicating a highly variable condition in his observation that "Now that's difficult. You have to describe it and you can't see it. All that you see was a muscle disorder." while for Maria "in my case it's [DM] quite invisible"

The visibility of DM, through the eye of the clinic or expert family member was also evoked. Maria described how "she [mother] used to notice things with me that I didn't notice myself. She said 'you're walking funny, what's the matter with you? And I'd say, 'oh, I didn't notice'. And then I had an assessment with Dr X to assess me for disability living allowance and he said, 'oh, you've got a limp'. I said 'I haven't'. He said 'you've got a slight limp on this side'." The mysterious nature of DM was alluded to in terms of how it might be affecting parts of the body not obvious to the person and where, as 47 year old Daniel reflected "What's going on inside is of more concern really."

DM was narrated in colourful and sometimes sinister language as an agent in its own right rather than as a phenotype of a faulty gene that was part of biological make up. It was typically narrated through accounts of everyday life where it functioned as a disruptive force, preventing or altering the normal course of events. For Edna "It's a nuisance I know that much because I can't lift anything heavy. I can't open jars or tins sometimes, you know." Lack of visibility, combined with accounts of unpredictability and uncertainty reinforced the innate mystery of DM.

Accounts of how behaviour in everyday situations had changed conveyed the threat to identity associated with a diagnosis of DM. They related to the person's behaviour and the behaviour of people around them. These narratives emphasised the way the person had been and contrasted it with the changes DM had brought.

> *I wouldn't; like I used to give him a kiss in the morning, no not any more. I wouldn't even let him touch me, and that was weird because I'm more like very ...Touchy feely kind of person.*
>
> *Louisa aged 39*

Kirmayer (2000, p. 155) wrote of how narratives can be fragmented and where experiences such as illness or distress "may undermine our efforts at self-construction". The use of powerful metaphors can illuminate narrative meaning in an account that is as yet unresolved. He asserts that, "Metaphor therefore occupies an intermediate ground between embodied experience and the overarching narrative structures of plots, myths and ideologies" (2000, p.155). The juxtaposition of an essential nature with a new and unresolved self gives a representation to the struggle to incorporate a new biological version of the self into the valued previous one.

Searching for knowledge

> *And if you don't know you can't ask.*
>
> *Simon aged 54*

The narration of DM in terms of the velocity of its unpredictable physical deterioration was mirrored in a process of adaptation, which was also ongoing. The search for knowledge was common to all of the accounts, regardless of how the diagnosis had been made. Many of these accounts were in the form of moral tales, indicating how good and bad practice had helped or hindered progress. The medical profession was the focus for some of these accounts. Particular narrative emphasis

was given to professionals who did not acknowledge their lack of awareness of the condition, or whose knowledge was limited and "You know, they don't know enough about it and they can get you more confused..." These professionals were generally differentiated from the DM geneticists whose expertise and communication skills were valued, although suggestions for better practice were given here also. The internet, the MDSG and specialist books were mentioned as common sources of knowledge. The narratives drew attention to the resource, the response it evoked, and the temporal aspect of readiness to absorb information.

> *I mean the first thing I did was ring the support group because I worked in the voluntary sector so I knew about medical support groups.*
>
> *Daniel aged 47*

The framing of the account in terms of what could be improved allowed the personal experiences of the family members to emerge and gave a context to why particular ways and types of information were privileged. Information such as books and leaflets that could be used when appropriate was frequently cited as useful.

> *...it would be nice if they could, you know, they can diagnose myotonia dystrophy. Here's three little booklets, that tells you everything you need to know. ..It tells you all about your benefits you can claim, all the help you can get, financially etc, etc.*
>
> *Emily aged 48*

Official letters from the geneticist were useful to provide information about the condition and also to validate it. This linked with the visibility of the condition and the gradual deterioration associated with DM. Again the language of detectives and crime was invoked as family members spoke of not being believed, especially in work, where provision of material evidence was necessary.

> *...and I had to get letters from genetics that is to prove that I actually had this condition...I've proved it. I've got consultant's letters, what more do you need.*
>
> *Maria aged 46*

The burden of proof invariably lay with the person with DM, but acknowledgement of the condition was seen as a mixed blessing. Gwyn (2002, p. 163) asserts, "Hanging over every patient is the potential accusation of malingering, resulting in the obligation to prove that the malady is not contrived..."

> *I can't explain what it is because I didn't even know myself. But they won't accept that. They won't accept that you don't know what it is. You are supposed to know.*
>
> *Louisa aged 39*

I think you're discriminated against because of it.

Simon aged 54

Having the condition was differentiated from knowing about it through accounts of frustration with the lack of concrete knowledge available.

> *Despite the condition being sort of recognised, very little is known about it really, or at least as far as I can tell.*
>
> *Daniel aged 47*

> *You understand what it is but you don't understand how it's going to affect you.*
>
> *Elizabeth aged 30*

The narratives also performed the function of making visible the dangers inherent in seemingly helpful situations.

> *It is like when I had my cataracts done private ...and the surgeon had the brochure [on DM and anaesthesia] and was reading it prior to me just going in and I thought I hope to God he's read that before I have had it done.*
>
> *Marion aged 60*

Family members related the constant negotiation of situations in relation to disclosure of DM and how their expertise about DM had developed over time although this paradoxically led to the conclusion that no two situations were ever the same, rather than the building up of a composite picture of DM.

The need for education at all levels formed the basis of many accounts, ranging from personal stories of how knowledge was necessary, to broader narratives of the importance of as many people as possible, particularly professionals, to be aware of DM. This was accomplished by accounts of helping in medical student training and clinics. Harry described how "There's a world book of knowledge...Well I'm in there." and Michael remembered how "I had a letter from one of the doctors that examined me asking if they could put a picture in a medical book."

Two types of accounts related to this educational role and where they differed was in the agency of the person with DM. In the first, described above, there was a stated willingness to help in order that "as many people as possible know about DM". The second scenario referred to situations where a person with DM became the focus of ignorance or consternation, and subsequently objects of instruction to others.

Although both situations might seem to accomplish the same aim there was a distinction made in the way the narratives were told.

> *...going in for my pacemaker. I was sitting there, this doctor came, well he had a white coat and he had a thing there, I thought he was a doctor... he said 'are you the gentleman that's got this myotonic dystrophy' and I said 'yeah'. He said 'oh, can we do a few tests on you'?*
>
> *Gareth aged 58*

Passivity further reinforced the lack of agency associated with having DM. This was apparent also in accounts of going to clinic and having unsuspected problems revealed. The DM clinic was the site of narratives relating to diagnosis, communication and surveillance. It was distinguished from other medical settings by the way family members constructed it as a source of expertise but also ambivalence. This was particularly true of stories of communication where the opportunity to talk to an expert was valued, but the frustration of not being given sufficient information about intervention was also recounted. Acceptance of the way the DM clinic staff monitored change varied from feelings of apprehension to a more prosaic view of the clinic as a place of routine checks that were just another aspect of having DM. An interpretation of the clinic as either a place where aspects of the physical self were revealed or as a place where doctors monitored DM for their own interest highlighted the importance of agency. Illustrations and accounts of good practice emphasised the cooperative nature of the relationship between the person and the clinic. For Emily, the geneticist "knows, she knows how to look after me. I think she does a damn good job...And she does talk to Alex [son with congenital DM]...And I may have to answer the question for Alex but she does try."

Situations where the geneticists had acted as an advocate and had listened were granted special status and contrasted with memories of controversial encounters where clinicians had not listened. Louisa highlighted the potential complexity of a clinic appointment by narrating how her husband always came into her appointment so she didn't talk of the issues that most troubled her in order to protect him. Daniel spoke emotionally of difficulties he and his wife had faced a decade before, in trying to persuade geneticists to test their young children for DM. His recollection of how "They never asked if we would still love them" reinforced a lack of agency and the need to take the family perspective into account.

Researching knowledge

It gives us hope.

Emily aged 48

The search for a gene and ultimately a cure has been represented in popular culture as a quest narrative where obstacles are overcome in order to reach an ultimate goal. Family members did not talk at great length about the discovery of the DM gene, although its translation into the availability of a definitive diagnostic blood test was narrated as a turning point in vivid and detailed accounts of diagnosis. Where the gene discovery was recollected it was a quest that was referred to in terms of a distant hope for the future. This was not seen as relevant for themselves but for the younger generations of their family. Michael remembered how "they [researchers] turned around in the hospital and said they had found the gene now and when [daughter] was older then they might be able to relieve some of the symptoms". Maria referred to gene discovery as "a great success" but "from my point of view it can't help me".

I've given quite a lot you know but if it is benefiting somebody else, not benefiting myself, then you know, look to the future, then it's worth doing it.

Leo aged 61

The narratives referred to cooperation with medical and scientific researchers who were part of the same journey of discovery as the family members themselves as "They're [researchers] finding out as they go along aren't they really?" Family members spoke about how they were willing to help with research and gave accounts of research in which they had participated, but were realistic about the timeframe for any advances. This pragmatism was narrated using temporal frames of reference with qualifying clauses such as "too late for me, but for my son, fabulous." The application of time to research was stressed in terms of how soon a major advance would have to be made in order to help the next generation.

I said to him [medical researcher], as soon as he come through the door, I'll give you all the time you need, I'll tell you anything, but you've got to find me a cure for this...you've got five years because my son was eleven then...I said you've got five years...because they said sixteen was about the age I'm getting him tested...So I said you've got five years to do something.

Louisa aged 39

Simon spoke of his hopes that "She's [grand-daughter] a 12 year old now but let's hope they can find something in the next five, six years for her."

DM was repeatedly narrated as a dynamic agent of change, rather than as a fixed concept encapsulated in a diagnostic label. This dynamism and potential impact on the next generation was also being documented and monitored. Hope, as an abstract concept was associated with research and this was given meaning by specific accounts of whom the research might help. Bakhtin (1987, p. 95) wrote, "both the composition, and, in particular, the style of the utterance depend on those to whom the utterance is addressed, how the speaker senses and imagines his addressees, and the force of their effect on the utterance". The information sheets sent to the families stated the purpose of the interviews as researching the impact of the DM gene discovery. The narratives chosen by family members did not foreground scientific research or genetic advance, with the exception of Emily's husband who emphasised his scientific knowledge. His account also framed the temporal aspect of a cure where "In America with mice, that had got myotonic dystrophy...They found that they can switch the defective gene on and off with antibiotics...It's probably ten years before anything may come through."

Genetic language was invoked to demonstrate competence in situations where there was felt to be insufficient or censoring of information from professionals. Family members spoke of their interest in research generally as a motivating factor, but also their co-operation and willingness to help wherever possible, without expecting any personal gain. This was made explicit in Daniel's interview where he said "I 'm quite happy to talk to you about it (inaudible) ... ... I wouldn't bore a neighbour or something about it... I mean I prefer to be reactive in this conversation rather than too pro because I would want you to have what you wanted."

The interviewer represents an audience and a communicator of shared meaning from the interview to other potential audiences (Bakhtin 1981). An interpretation of the interviews could therefore include the shaping of their content by the families towards these other audiences, notably the doctors and the clinic referred to in the information sheet. This infers that topics chosen, and the narrative devices chosen to communicate them, offered an alternative classification of DM as a medical and molecular condition to one with profound effects on everyday family life, characterised by living with uncertainty and the need to regain agency.

communication in some cases over medical tests, which were interpreted by family members as having predictive value although from the practitioner's point of view they may have been routine. Louisa expressed her anxiety about a clinic test where "I've heard nothing, nothing. I don't know whether things are alright or whether it's bad."

Again this related directly to everyday function and accounts were given of dilemmas over whether to make particular plans for the immediate and long-term future. These were contrasted with accounts of the difference that being able to talk to a professional who understood the condition could make. In these cases narratives focused primarily on the DM clinic, but also in two interviews with stories of home visits by a specialist genetic nurse. Taking time to talk and having expert knowledge were viewed positively.

Differing interpretations and experiences of the condition within families also contributed to uncertainty. Alan struggled with the fact "...my father never said anything about any pains you know anything like that? Like I said, I ache constantly, every time, every day, all the time." The knowledge that DM was unpredictable and could manifest differently within a family added to the frustration of trying to retrieve agency. For Louisa "Because it's progressive and like my sister's in a wheelchair and, and my brother's in a wheelchair and I'm thinking I know I'll be heading that way. Obviously because if they have, I probably will..."

Disclosure

> *Then my friend when eventually it was diagnosed and she said "Thank God", she said "He's like Paddington Bear because now he's got a label"*
>
> *Marion talking about her son Steve*

The contingent nature of disclosure about DM diagnosis was a theme across the interviews. Many family members told stories of situations where use of the diagnostic label had both positive and negative implications. These stories were primarily structured as accounts of motives, with choices over disclosure being explained or justified.

> *Once they labelled her [grandchild] with that it was like well she was never going to get into that mainstream. And I don't say she was just left. I don't mean that. But they didn't work with her as well because they knew that she*

Agency and Isolation

So it affected me a lot because I felt it was responsible for that and I was really angry then, I thought it was unfair.

Maria aged 46

Family members distinguished between stages of coming to terms with DM and coping with the physical changes caused by it. Words associated with blame were used to describe the inheritance risks. When discussing how narratives are used to make sense of events Ricoeur (1984, cited in Smith 2008 p. 115) stresses the central function of agency as a way of communicating the interviewee's role or lack of role and attests that "The converse of agency is suffering". This use of the word suffering also corresponded with Kleinman's (1988) analysis of chronic illness narratives. However there were differences in the way family members spoke of the personal meaning of DM to them. There were references to bad luck and unfairness, but also to relative fortune at being less affected than other people they knew with the condition. Maria spoke of DM being "hard luck really" and how "sometimes I think it's unfair. Why have I got to put up with this?" This contrasted with Gareth who expressed "I'm lucky in a way" and illustrated this with an example of "the good thing, it's not painful. That's the one thing, no pain with it. That's the good thing about it." Unfairness was associated with lack of agency and was reflected in use of the passive voice where DM was "put up with".

No-one understands what it is like unless they have it

Alan aged 33

The narratives reconstructed the difficulty of really knowing what it could be like to have DM. This was expressed even in families where the person interviewed had grown up with it, and served to reinforce the complexity behind a diagnostic label. Harry made an attempt to explain this complexity scientifically by referring to the number of repeats in his gene test result and how he had been "off the scale". However the accounts evoked isolation through the use of phrases and language conveying how it felt to be constantly misunderstood such as Rebecca explaining how "It [DM] makes you sound drunk."

Misunderstanding of DM was narrated across all areas of life and included medical personnel as well as family, friends and work colleagues. The desire to establish agency and some personal control over the future was also confounded by lack of

would be going to a special needs school then. So in a way perhaps it's best not for a child or for the family to know.

Rachel, partner of Simon

Caroline related her son Brian's experience of mainstream school and how she told the head-teacher "I'll get somewhere else for him because you know, you're not doing anything for him. The child is stuck in a corner, you're just not interacting with him, it's not fair."

There were dilemmas over the personal gain of accessing resources and help through the recognition of DM by others, versus the difficulty of being defined purely by the diagnosis. The fluctuating nature of the condition and the way a personal philosophy for living with this uncertainty had been, or was in the process of being attained, was narrated. However this did not happen in isolation and the capacity of others to interpret DM for themselves and react accordingly was evoked in accounts of teachers treating children differently following diagnosis, or work situations where "Because actually the first time I mentioned it [DM] in work they [colleagues] said 'No, you haven't got that'."

Stories of the constant need to negotiate situations on an individual basis, and also to evaluate possible difficulties, served to further make visible the shifting assumptions with which family members negotiated everyday life. Even resources where the primary aim was help and support could not be accessed without consideration of potential hazards.

We went on a myotonic dystrophy support group but I don't think it's going to be a help at all...Because everybody was so different...there was somebody younger than Leo there and I mean, I thought, I've got to watch what I'm saying you know, I couldn't tell her how things are going to develop because it could be totally different, totally different. We've seen enough of it in the family with four of them with it, not one of them are the same you know?

Sarah, Leo's wife

The receiving, seeking and communication of information was a continuous process, according to the families, with changes in function leading to the need for more information and the need to adapt in everyday life. This management of knowledge was narrated in several accounts that dealt specifically with how and when to talk to children in the family about their parent's condition and also the potential implications of this for the child. Louisa articulated her worries about her son Luke

questioning DM in other family members and asking" will I have it? I know he's going to ask me, and I, I don't know what to do with that. Because I'm going to have to say well you might have it."

This very practical parental issue was related primarily through stories about relationships, although accounts also included reference to both positive and negative potential effects in everyday life. The placing of a child in appropriate education or access to specialist equipment and resources was weighed against potential stigmatising of the child by a label. The accounts used descriptions of what the child enjoyed doing and placed this against a scenario where knowledge of the condition might change the approach the child took to life and the wish that "... I want him to have a normal; as normal childhood as he can."

Fear of changing perception also related to affected parents, where the child might have to take on responsibility for a parent with disability. Michelle articulated her personal concern about having to use a wheelchair in the future but expressed concern also for the implications this would have for her daughter Stella. For Louisa her identity and role as a mother was closely bound up with fear that she might have passed the condition to her son and that her condition would lead to a role reversal where he would become the carer.

> *...if you've got a mother that you've got to push around in a wheelchair and stuff like that...Now when I see old parents and the kids have got to look after their parents and that...Because I feel like oh that'll be – that'll be me. And I feel so sorry for those kids.*

Stories of how she had seen his reaction to children with disabilities reinforced her ideas. Her earlier use of the word "surveillance" to describe how she and her husband constantly monitored their son related to his physical condition and to the emotional and social implications of telling him about DM.

The narratives all had a common temporal aspect of an optimal time, or at least a time by which it was necessary to tell a child about their risk of DM. This was associated with forming relationships. DM was specifically narrated as a genetic condition in relation to the future of the next generation and reproductive awareness. This was regardless of how positively or negatively DM was narrated in the interviews.

Caroline spoke of her teenage son Brian's wish to have a girlfriend and the challenge of encouraging his confidence in his abilities in the special needs training college he attended while also communicating a sense of responsibility.

> *He likes girls, different one every week. We said don't touch them, because I don't want you touching them, and somebody coming on the phone to me...He says... I haven't kissed anybody; I've hold their hands. Well that's alright then. You mustn't touch. (inaudible) because he's got to be told. He knows where babies come from*

This concern for telling children, and the dilemma of how and when to do it was also associated with extended family. Caroline and Rebecca described monitoring their nieces for signs of DM as they reached reproductive maturity. In some cases DM was recognised by one branch of the family but not engaged with by another. This did not necessarily lead to a fracturing of relationships and again negotiation of social and biological identities was narrated. Responsibility for informed reproductive decision-making in future generations was inferred in all of the accounts by the way in which stories moved to a future not related solely to personal change but also the implications for younger members of the family.

Reconfiguration

> *I didn't want to pass it on. I thought, it stops with me.*
>
> *Gareth aged 58*

Both men and women in the family interviews narrated the altering of reproductive possibilities. Narratives highlighted dilemmas and explained past actions. Of the five men diagnosed with DM who did not have children, two were in long established relationships and both spoke of the enormity of their decision not to have children in order that the condition was not passed on. For Gareth and his wife Sandra "Not to have children was our worst decision really."

Michael and Alan discussed their social responsibility for passing on and preserving the family name but for whom a greater responsibility for not passing on the gene had been taken. Paradoxically the accounts of the choices made highlighted agency, although neither of the available choices were desirable. The sadness expressed by Michael where "That's what I missed most, not having a son to keep the men going" was echoed by Alan, "...so I'm the last in my line basically so I'd like to have kids to carry on the line but it's not going to happen because it's too selfish. I mean I'm so

upset about not being able to have kids." Michael and his wife Sarah have a daughter and grandchildren, and had resisted pressure from family to have any form of testing in pregnancy as they were committed to having a child. The diagnostic blood test was not available at that time but there was a confirmed family history and both Michael and Sarah were familiar with the manifestations of DM. However after the birth of their daughter, whose photographs were displayed all around the room, they decided they did not want to expand their family, as the risk of DM seemed too great.

The work of parenthood was visible in the accounts from parents diagnosed with DM, and those who were unaffected. Narratives of parenthood included active reproductive decision-making resulting in making a choice not to have children, and also in the experiences of traumatic reproductive events. Maria recalled how she "worried about it [DM], because I was thinking well this could really affect the baby and he [ex-spouse] said he was OK with it and willing to take the risk if I was OK. I said 'no I'm not...I'm not OK you know I have got this condition and I may pass it on or may not' ...but – you know – he just wanted a baby and he wanted his own baby and he refused to adopt or anything like that and sadly it didn't happen." Anna was diagnosed when she lived abroad and remembers her doctor saying telling her she should be sterilised as she had a 95% [incorrect] risk of passing on DM. In reflecting how she hadn't wanted to "bring that sort of suffering into the world" there was also regret where "Maybe it was a good thing. But I also feel a loss about it".

The narrating of these events indicated their centrality to personal identity. In choosing to talk about their choice not to be a father or the trauma of miscarriage the conflicting desire and fear of being a parent became visible.

If she [daughter Stella] had a child that didn't have it the chain could break
Michelle aged 37

The mothers interviewed spoke of their worries for their children and their feelings of guilt at having passed on the condition. There was reference to possible termination of pregnancies in two situations, one of which was offered medically and the other suggested by extended family. The influence of family was visible, either directly or indirectly through their experiences.

...before we got married I got told [by her parents], "Are you sure you're doing – you're doing the right thing. If you have any children, they might be affected"...we were offered a termination when I was pregnant...And we said "no, it took us long enough to get there, we'll just cope with whatever comes".

Sarah, Michaels' wife

Caroline spoke of the pressure from extended family to terminate a second pregnancy; whereas her feelings were that her experience of having DM and having a first child with DM would prepare her better.

I was worried. [Mother-in-law] told me to get rid of it. But if it was the same as Brian [elder son with DM] at least I know what to do. Where to go...And she told me to get rid of it but... it wasn't in my nature to do it.

This pregnancy resulted in a son who does not have DM.

Joe, a 58 year old man diagnosed with DM, also referred to pre-natal intervention in his family where "They took a gene out of her [sister] womb because that's what they didn't do with the first baby..."

Although one of the key features of gene discovery was the introduction of the diagnostic DM gene test enabling individuals to ascertain their DM status without the involvement of other family members, the interviews illustrated the influence of family perceptions and views on reproductive decision-making. As most of the women were unaware of their status when they had their children they did not speak of reproductive choice in the same way as the men. Louisa, who does not yet know her child's diagnostic status, was extremely upset and emotional talking about all aspects of family and reproduction and recalled how "When they confirmed it [DM] I made the decision then to be sterilised straight away".

Emily, whose nineteen year old Alex was born with congenital DM leading to her own diagnosis, reflected on the fact that by not knowing about her status she did not have to make decisions about her son, as "I wouldn't change it now. But if I'd known I don't know if I would have done."

Interpretive repertoires and Performance

Culturally available resources for narrative construction of experiences were as a patient, a disabled person (as opposed to having a disability) or a person who was living a "normal" life. Narrative identity as an ill or sick person was rejected explicitly in several interview accounts, and there was ambivalence about the perception of disability.

There was a limited range of repertoires available through which to narrate experience and this was reflected by the rejection of having to take on a defining role of being a patient. Roles of everyday life were valued and the frustration of trying to find the resources necessary to accomplish them without relinquishing the status of "normality" was referred to repeatedly. This extended to partners who spoke of how difficult it was to maintain everyday life whilst having to portray worst-case scenarios in order to receive help. This placing of identity in restricted genres or repertoires again emphasised the threat to agency and autonomy implicit not solely in a diagnosis of incurable and degenerative illness but in the resulting limitations to living a desired life. The wheelchair was symbolic in several accounts of the relinquishing of a previous identity. There was one account that differed and where agency extended to how equipment such as a wheelchair could be used to restore a valued activity. It was the association between the activity and the object, rather than the symbolic nature of the object, which restored agency. Michael described how he had used disability equipment from the local council on a shopping visit and "I had the scooter. It was great. First time I'd been in [the city] – having a ride round because I couldn't walk, 'cos we used to walk round".

Being able to access resources and assistance when necessary, without being defined by one role, was valued and appeared in narratives about adapted transport for holidays, the availability of hand rails in entertainment venues and the sensitivity of a friend in recognising that the situations where help was needed varied.

The language of the interviews contrasted markedly with their physical performance. Michelle spoke of how this reflected her experience in everyday life where her physical appearance and ability affected her social interactions.

Every day that goes wrong inside, your legs are weak, your hands are weak, your face... the muscles in your face goes, you can't really smile, so you can't really be friendly.

In their narratives family members acknowledged the impact of excessive sleepiness, which is documented in medical classifications of DM, but challenged perceptions of associated apathy by placing it in the context of everyday life and illustrating the effort required to accomplish daily activities.

All of us suffer from it is the get up and go feeling? You haven't got it. You just haven't got the incentive, not incentive (inaudible). You just can't be bothered and we're all like that...If I want to do something I force myself to do it (inaudible). I don't know whether I want to go to the club or not. But I think well if I don't go I'll be in for the rest of the week.

Simon aged 54

By separating out the language from the performative aspects of the interviews it was possible, through the linguistic choices and narrative context, to see more clearly how the active work of reaching a personal meaning and establishing some agency were challenges to the physical implications of decreased ability. The language was significantly more powerful than the appearance of the person using it would suggest. In this way the narratives and language chosen by family members privileged activity and motivation over passivity and apathy, whilst recognising that these embodied the challenges of DM to them on a personal level in everyday life.

Searching for meaning

It's a journey

Emily aged 48

There were many stories of learning to live with DM. These were constructed using dynamic and powerful language and echoed the powerful representations of DM. They stressed the insights achieved through the process of engaging with the implications of DM. This too was seen as an active rather than a static achievement with phrases such as "There's so much I have to do to prepare for the future" revealing the ongoing adaptation necessary. Diagnosis functioned as a key and symbolic reference point in the shaping of these narratives, but in re-telling the story in a different form it marked the beginning of a new era in the life of the narrator. Metaphoric descriptions such as "It's a learning curve" suggested accrued effort resulting in the attainment of the necessary knowledge, skills and agency to manage life with DM.

That's the way it is

Simon aged 54

Many family members related DM as something that could not be changed. However the way DM could be managed or integrated as an attitude added another dimension to the uncertainty inherent in the DM diagnosis.

> *But it was the Saturday when I first heard, it took me a while to get over it, but now I just accept, well, I've got the condition I've got to do the best I can.*
>
> *Maria aged 46*

Family members constructed personal and emotional accounts of living with uncertainty and gradual deterioration. These accounts differed in ways of dealing with these issues but shared an awareness of either an attitude or meaning which had been formulated over time, or a need to gain some degree of understanding over the introduction of DM onto a life course. A change to the use of the second person in the accounts conveyed where some degree of personal meaning had been achieved.

> *It's difficult because we have coped for so long on our own you know. You just get on with life.*
>
> *Marion aged 60*

Awareness of the need to integrate personal meaning did not necessarily result in everyday life becoming easier. This was explicitly conveyed in Michelle's account where she spoke of how "I can't do anything about it so I've just got to get on with it. You can't just sit in your bed or lie in your bed and cry about it because it's not going to go away..." The accounts communicated personal meaning as the need to establish an attitude or assert control over a condition that still remained variable and unpredictable. Use of moral tales, in the form of life lessons conveyed instruction or learning.

> *But you accept it more. I know that the first two years is terrible. I've had it four years now. I think I've come to terms on what I can do and what I can't do. Whatever comes you've got to deal with it as it comes. If you can cope with it better it comes on slowly.*
>
> *Simon aged 54*

Family members narrated personal meaning as a turning point in their biographies. DM continued to be distinguished by an independent trajectory but assertion of a personal strategy or outlook reintroduced a degree of agency.

Narratives given by the family members and their partners stressed the significance of contingency and the necessity of ongoing adaptation in everyday life. The

accounts utilised the second person to convey the unpredictability of DM and how personal resources for dealing with it were also subject to fluctuation. This constructed an overall perspective on an ongoing narrative, and also a philosophical approach that resonated beyond the diagnosis and was made accessible to a wider audience. It was part of the repertoire of personal meaning that was present in all of the interviews; even those that revealed unresolved issues relating to acceptance of DM. All of the interviews were characterised by an active working towards meaning rather than a passive acceptance. Narratives of good days and bad days conveyed the effort required to accomplish everyday life with DM. Although the phrase "there's nothing you can do about it" appeared with regularity, it was accompanied by statements indicating that acceptance was only part of the necessary perspective. Maria and Edna referred to their ability to "have a laugh" as a way of indicating normality and control. In other interviews everyday activities such as going into town shopping or seeing friends accomplished balance.

The accomplishment of these activities required consideration of resources, both environmental and personal, in many cases. The immediate terrain required negotiation as falls and unsteadiness were frequently narrated. Sarah, Leo's wife, spoke highly of the advice Leo had received from health professionals about prioritising activities and conserving energy. The physical expenditure of energy was weighed against the overall benefit of an activity, as Simon conveyed in his account of walking uphill to his club once a week, despite the resulting tiredness, as it enabled him to socialise.

Narratives communicating the ups and downs of everyday life gave insight into the reality of living with the condition. Anna explained how DM had been in the background until she began to have difficulty walking. The uncertainty, where "You don't know what the future's going to hold. You know it's going to get worse" was juxtaposed with having to "face up to it". Anna and her husband both expressed the value of living with DM on a daily basis and coping with situations as they arose.

DM threatened self-perception and roles and relationships within the family. DM was narrated primarily through its ability to effect change over time, rather than in terms of its genetic features. This extended beyond the time-span of the person narrating the story to the future of their family.

...it breaks my heart that I can't play football with my son.

Daniel aged 47

...our son, looking back he had to grow up quicker because I depended on him a lot which I should have turned to his father ...

Sarah, Leo's wife

Mum was the man of the family.

Alan aged 33

The narration, through roles of everyday life, of the ability of DM to influence and change, again reinforced a key theme of how it impacted on families.

Biography

So it's been all those years you know. Some good times and some bad times.

Gareth aged 58

The evaluative function of narratives, and the way they reflect a particular set of circumstances, was apparent in the temporal aspect of the interviews which encompassed past, present and future, and that at times bound all of these together in a meaningful way. This functioned to illustrate the contingent nature of coping. Family members narrated how personal meaning was attained by integrating unpredictability into an approach to everyday life, and by accepting that the condition manifested itself in ways that could not be controlled.

Louisa's account moved between the past, present and future as she narrated the loss of her role as a loving daughter in the past, a loving wife in the present and the possible loss of her role as a loving mother in the future. Her use of the phrase "it's haunting all the time" to describe her continual struggle to come to terms with DM had particular resonance with the stories she told of her changed behaviour, and "that's why I'm not coping; I know I'm not coping because I'm doing things like that...And that's not my nature."

The temporal aspect served to map the trajectory of the condition alongside the trajectory of the person's day- to- day life. This enabled a picture over time to emerge that was characterised by the attitude of the person rather than by the deterioration or loss of physical function. The use of phrases such as "you've got it so carry on, we just laugh and joke about it and as we saw things happening we changed them" placed agency back with the person rather than with the agent of

change, which was DM. There were narratives that referred to this differently, with agency being the defining element. Michelle described how "There are days when I'm happy and nothing goes wrong, and then I have days when everything goes wrong." She repeatedly expressed a lack of control throughout the interview, and although many of the phrases used were similar to other accounts such as "There's nothing I can do about it...it's just one of those things", the evaluative aspect of her narrative highlighted the sense of unresolved agency and control.

Stories of the past where "I used to do cross country running, play cricket, rugby, football, swim, walk..." enabled a previous version of the person to be validated and evoked alternative roles and selves to the present. The narratives illustrated the life that might have been if the diagnosis of DM had not been made. People who were recently diagnosed or struggling to come to terms with their diagnosis spoke of future roles, which they were relinquishing. There was a continuum of experience from the ongoing dilemma of negotiating ordinary life and life with DM, and being perceived as a disabled person by accepting help.

This reflective perspective included details of what had been achieved and the effort it had required. Evaluation stressed the active effort involved in seemingly passive phrases such as "it is as it is", "it's inevitable" by juxtaposing them with descriptive phrases such as "I've learnt to live with it" and "I won't give in." References to "good days and bad days" reflected the temporal aspects of living with DM and how "Time is placed in a personal history" (Coffey and Atkinson 1996, p. 68).

Key turning points in personal biography such as diagnosis or reproductive decision-making were narrated in terms of potential impact. Family members constructed accounts around the language of loss to speak of their changing physical condition. This language of loss extended to possible future loss, particularly of valued roles or anticipated life stages such as retirement where "him and me have always said, oh we'll retire early. I know I won't the way I am. If I'm in a wheelchair or can't talk (inaudible) what's the point of retiring early?"

We used to go for walks and walks, didn't we, miles we used to walk, didn't we?

Michael aged 50 and his wife Sarah

Accounts of how roles of everyday life had changed were narrated to convey the implications of DM. Unpredictability was a theme affecting all areas of life. This was communicated through contrastive rhetoric. The narrating of how life used to be was integral to these stories and implicit within this, what life could have been. The phrase "used to do" appeared frequently throughout the interviews. It was used in accounts of everyday tasks that could no longer be achieved, and functioned as a temporal comparison. This comparison was used to describe relationships, previous interpretation of signs and symptoms, accomplishment of roles, and changes observed and recorded in the DM clinic. As such it functioned as a reflection of change in the condition but also was illustrative of a before and after. Through her account of how her ex-spouse refused to acknowledge her diagnosis of DM and the subsequent physical and emotional pain of a miscarriage, attributed with hindsight to DM, Maria made visible once possible and intended roles as a mother and wife.

By contrasting through narrative what had been accomplished in the past and what was possible now, family members communicated their gradual deterioration in function. This was accomplished through the telling of stories located both in the everyday and in the clinic. The difference between these stories lay in the way the information was made visible in the clinic by experts revealing hitherto unknown or unseen aspects of degeneration through tests and measurements, and the practical resources based in everyday life, which enabled family members to narrate their own changes. The significance of monitoring change was inferred by the frequency with which it was related in the interviews. However the way the clinic was a place where knowledge lay in the hands of others was apparent in the degree of anticipation that was expressed about attending. For some it was a welcome opportunity to "keep an eye" on things but for others it was a place where "they always find out something".

Narrating what a person used to be able to do communicated the degree to which DM affects function and enabled the narration of other possible selves, consistent with Ricoeur's (cited in Smith 2008, p. 247) assertion that "Subjects recognize themselves in the stories they tell about themselves". Stories of holidays taken, sports played,

and previous places of employment lent a temporal aspect to the trajectory of a person's life, not a one-dimensional trajectory of living with DM. This extended to actual aspects of the self and also to possible roles in the future.

Narrating future roles allowed possibilities that were no longer viable to be witnessed and recognised. The emotional language used to describe coming to terms with loss of roles implied how integral this was to the experience of having DM. This was also the case for partners, who spoke of how their own lives had changed. Simon's partner Rachel remembered how, on hearing of his diagnosis of DM and that of their daughter Elizabeth "The first thing I thought of is 'Oh god am I ever going to have grand-children' while Sarah, Michael's wife reflected that "I think my life has changed as well 'cos there's a lot of things I can't do anymore and I haven't got the condition." When Rachel was asked for her thoughts about DM she replied, "You're the first person that asked me that. 'Cos I think well everybody do ask how they [Simon, Elizabeth and Clara] are feeling. Nobody thinks to ask how I feel". The valuable contribution of partners to the interviews was unexpected, and in most cases appeared to be unanticipated by them, as they moved from doing other activities to join in and offer their perspectives.

Contrastive rhetoric illustrated aspects of change in accounts of how things used to be. This functioned to compare past with present and to make previous and now unseen past selves visible. It showed how the imagined future had to be reconsidered and how taken for granted roles within the repertoire of everyday life were threatened or in need of review.

Holidays were a recurrent topic across the interviews. Initially this seemed to be a way of accomplishing small talk but analysis of emergent themes from the early interviews indicated its significance as a way of marking temporal points on the trajectory of DM and the trajectory of everyday life. Sophie, who was physically extremely frail, spoke of how "You shouldn't give in. I'm a bit stubborn like that. It's a good thing I think 'cos if you give into it (inaudible) just forget it. That's why I (inaudible). It depends on how badly you're affected obviously." She returned several times to her passion for travel and how she had found companies that accommodated disability. Emily's stories of holidays with her husband and son Alex,

who has congenital DM, were typical of many accounts in the way they were structured around accomplishing activities "before whatever [aspect of DM] kicks in." Her husband spoke of how "We did try and get things done, you know like, holidays" as did Rebecca's husband Mark who described how they could no longer fly but had found appropriate holidays in this country.

The incorporation of the trajectory of DM into a personal life trajectory was also narrated as acceptance expressed by Maria's belief that "I mean you can't sit around and moan about it 'cos you've got it and you're growing to grow old with it, you know, hopefully."

Summary

I said I got a piece of her life now.
Louisa, speaking of her sister who is severely affected by DM

Immediacy and clarity of recall characterised narratives of the impact of DM, whether by a recent and unanticipated diagnosis, or the confirmation of a personal diagnosis within a family with a DM history. Family members used vivid language to describe DM and portrayed DM as having its own trajectory that was now travelling alongside the future trajectory of both family and individual life. Phrases such as "carried" depicted it as a burden rather than an integral part of oneself. DM as a transformative agent acted upon self- identity and roles and relationships within the family.

Family members structured their accounts around the impact of DM on the roles and activities of daily life. They narrated how the new reconfiguration of the body through DM diagnosis changed perceptions of the physical self and changes in how family relationships were viewed. Childhood memories, expectations and experiences of parenthood, and anticipation of roles in the future such as retirement, were transformed by this new knowledge.

Accounts of work, parenting, holidays, interests and hobbies illustrated how, although the concept of DM was that of an unpredictable "other", its manifestation was rooted in everyday life and was integral to experience of everyday life. Family members measured change through their ability to carry out familiar activities and

narrated how these familiar activities were rendered unfamiliar or impossible due to physical changes.

Stories of everyday life served to illustrate the contrast between a former and a present self, as a way of measuring change, and to communicate how a diagnosis of DM did not define a person. Instead, the narrating of stories within a common repertoire of accomplishing everyday life situated DM as a challenge to, rather than an alternative way of living a life. In their accounts family members conveyed the ambivalence and constant negotiation of identity where deteriorating function necessitated practical help but where there was reluctance to adopt the label of patient. How others perceived DM was central to these accounts, as they potentially had influence over the possibility of continuing with a valued activity. Narratives of employment, where colleagues and superiors were central to the access of resources and opportunities, illustrated the difficulty of validating a condition that was variable in manifestation and visibility. Joe and Simon spoke of difficulties trying to find a job now that their previous skills were lost due to deteriorating muscle function, but where they were not equipped to work in a different environment. Cataracts made it impossible for Joe to continue as a driver but "The only job I can do is an office job and I've never worked in an office in my life."

Narratives of frustration characterised the difficulty in giving an exact prognosis. Again these narratives stressed the willingness of the family members to continue with valued activities. Autonomy and agency in deciding what help was necessary, and being able to access it were narrated as ideal but challenging to attain. Moral tales demonstrated how autonomy in accessing help without relinquishing of identity was privileged.

Partners played a central role in the interviews. The difficulties in planning ahead or accomplishing everyday life when a partner was unwilling to accept the diagnosis contrasted with narratives where there was acceptance of DM. Emily and her husband repeatedly narrated situations where he had demonstrated competence, often through challenging ignorance of DM or highlighting his medical knowledge in clinical situations. She summarised this assertive attitude, saying "I mean, I'm lucky because he can take it. Another man well, might, might not be able to take it. He's

going to take it on. But it's not going to be easy for him, because things are getting worse. No good denying is it." Where partners had reached a common understanding of the meaning of DM for their future this was integrated into their accounts of coping with everyday life.

The language used by family members to construct their narrative accounts of DM evoked strength, awareness and determination in a way that physical appearance and impaired speech could not convey. They narrated a corresponding attitude of coping with the unpredictability and deterioration of DM by changing and adapting, and by integrating those aspects of uncertainty into a personal outlook that was hallmarked by contingency and resilience.

Chapter Seven

Everyday lives, Genetic tomorrows

Introduction

This thesis explored the impact of the 1992 DM gene discovery through analysis of the accounts of the local scientific team, the perspectives of families attending the DM clinic, and documentary analysis of the DM medical record archive. Narrative analysis revealed the use of an emotional register highlighting the significance of relationships and dynamics in the everyday work of the scientific team, and the impact of discovery on subsequent careers. Family members gave accounts that were distinguished by vocabularies of strength, giving insight into the ongoing challenges of incorporating the DM diagnosis into everyday life. DM as a genetic condition emerged in relation to reproductive risk for the next generation. The scientists vividly reconstructed past memories of gene discovery, contrasting with the family accounts where gene discovery emerged within a narrative of hope and a willingness to cooperate with research for the benefit of future generations.

The medical record archive traced all aspects of clinical management and research into DM, from the first referral marking the beginnings of research grounded in relationships between clinicians, families and the laboratory, to the embedded practices that characterise the work of the contemporary DM clinic. The gene test was visible as a turning point. The medical records showed how the stress of living with diagnostic uncertainty was resolved after gene discovery with the advent of the gene test, but how management of uncertainty is now related to prognosis.

These findings are examined in the context of other relevant work and the potential for future research considered. Firstly the use of narrative is examined as a way of illuminating less visible aspects of landmark scientific research into DM, and the experiences of family members with DM. Agency is a key aspect of analysis in both the scientists' and the family interviews and its significance is discussed in relation to revealing additional perspectives on living and working with DM.

The second focus is on the striking emotional register apparent in the scientists' interviews, and the significance of this in relation to the sociology of scientific discovery in the context of everyday work. Thirdly, the translation of the scientific

discovery into a definitive diagnostic DM blood test is discussed in terms of the DM clinic, perceptions of kinship, the relationship between research and service, and the incremental nature of scientific and clinical progress. The implications of assessing the impact of scientific and clinical developments through a focus on families in addition to professionals are discussed in relation to clinical genetic practice and to sociological research into medical genetics.

Finally the potential for narrative analysis to illuminate less accessible aspects of experience is considered. Through focus on the narrative competence of the actors themselves and preservation of context the personal meaning emerges.

Expression

> *That's what's the worst thing. It's the worrying people. I can see people worrying. I can see it in their face. Even people who aren't related to me...In their mind or their subconscious somewhere they're like, oh it's a terrible thing to have. And people are really draining and I'm thinking, I haven't hurt anybody, I'm just, well I'm just going through life the best I can.*
>
> *Alan DM family interview*

The seminal work of Bury, Charmaz and Williams introduced a narrative analytic perspective that focused "directly upon peoples' subjective experiences within the contexts of their everyday lives" (Lawton 2003, p. 23). The impact of chronic illness on personal biography and relationships, with the self as well as with others, moved the context of evaluation from one of the "sick role" (Parsons, cited in Morgan et al. 1988, p. 45) to how individuals experience and perceive illness. Themes arising from the DM interviews included a process of attaining personal meaning, dealing with uncertainty, and changing roles and expectations. The expression of the DM phenotype varied, as did the expression of the personal meaning of DM through the family narratives. However, although their accounts had many features in common with narratives of chronic illness, the families did not define them in this way. Narrative was interpreted as part of an active process of meaning making, where moral tales, contrastive rhetoric and metaphor functioned to illustrate the trajectory of DM on the trajectory of personal biography. Through narrative family members constructed accounts that conveyed and shaped these experiences.

Families with DM constructed their accounts in the everyday and reclaimed their experience from a definition of disease to one characterised by uncertainty but

focused on accomplishing valued roles of everyday life. The focus on the everyday did not diminish the narrating of pain and distress that was integral to many of the family narratives, but served to contextualise the way a long term medical condition is bounded medically in relation to diagnosis and classification but is part of everyday life in practice. Analysis of how families who had been involved with the DM clinic over several generations interpreted the gene test, diagnosis and reproductive implications of DM highlighted the wider context of genetic practice and the influence of family experience and dynamics on decision-making. The primary focus across the narratives was the ongoing and varying challenges of accomplishing everyday life whilst acknowledging the significant and in some situations the overwhelming impact of the DM diagnosis. Families where there was previous familiarity with DM did not necessarily share the "biographical reinforcement" (Carricaburu and Pierret 1995, p. 65) noted in studies where existing morbidity had already altered expectations (Pound et al. 1998, cited in Lawton 2005). In the DM interviews men diagnosed with DM transmitted from their father particularly emphasised wanting to deal with it differently and more assertively. The variability of DM made it challenging for family members to learn their likely progression from other relatives. The influence of work colleagues and close relationships was narrated through all the DM interviews as a factor influencing ability to cope, echoing work by Pinder and Anderton (1995; 1989).

The use of metaphor in the family interviews constructed DM as mysterious, malign, unpredictable and having agency. Hallowell and Lawton (2002, p. 429) reported the use of metaphor to represent a genetic condition as being out of personal control in the way women compared their risk of ovarian cancer to a "time-bomb". The significance of the active properties attributed to the DM gene contrasted with the lack of agency that was perceived as one of the most distressing aspects of having the condition. This lack of agency was not expressed simply as a consequence of gradual loss of function but as a consequence of inability to attain information about the future or receiving inadequate information about health related procedures such as results from tests.

The expression of agency through metaphor highlighted its fundamental importance in coming to terms with the condition. Situations where agency was limited were

also expressed metaphorically. The observation that "metaphor is closely involved in structuring everyday thought and action, rather than merely being a decorative rhetorical device" (Gogorosi 2005, p. 300) was reflected in family accounts of living with DM and scientists' accounts of discovering the gene. Paradoxically the deterministic attributes suggested in the family metaphors for DM related to implications of DM in every day life that were partly a social consequence, such as difficulty accessing resources, rather than purely biological such as a measurable decrease in muscle function. The difficulty inherent in establishing DM prognosis is mirrored in the uncertainty and elusiveness associated with its metaphorical portrayal. This "constitutive role of metaphor, analogy, classification, narrative and genealogy" is described by Franklin (1995, p. 172) as "a cultural hermeneutics of knowledge" and gave insight into the impact of DM on family members through exploration of language and practices.

Interpretation of the scientists' accounts gave insight into the processes and personal meaning of gene discovery through analysis of the language and practices used to reconstruct events. Contemporary narrative reconstructions with the scientific team related to these dynamics and also the use of accounting devices such as luck and judgement as described by Gilbert and Mulkay (1984). In addition the scientists' accounts of gene isolation were embedded in relationships, dynamics and emotions. The interpretation of scientific culture has been a focus of sociological and historical interest. Scientists themselves have published work outside scientific academia but this has been related to either increasing popular understanding of scientific facts, offering personal scientific insights, or offering a personal perspective on a particular phase of research that captured the popular imagination such as the biography of Rosalind Franklin or Watson's "The Double Helix" (2004; 1996 [1968]).

In the scientists' accounts evaluation of the day-to-day processes underpinning discovery were clearly distinguished from evaluation of the scientific achievement of identifying the DM gene. Franzosi (1998, p. 534) observed how, in scientific writing, "the authors' direct intervention is minimal if not nonexistent"; while Woolgar (1981) speculated that the reason why scientists don't privilege reflection is so they can establish objectivity about their accomplishments. The scientists created accounts that gave insight into the process of scientific discovery through the

narrative structuring of plot and motive, and the use of narrative devices such as moral tales and accounting devices

Narratives of building models conveyed how the scientists carried out non-verbal activities "especially at the frontiers" (Gooding 1992, p. 66) in order to extend knowledge. The scientists highlighted the culture within which gene isolation was accomplished as creative and structured around problem solving. In recognising that "science is a form of culture with its own creeds, language, material practices, perceptions, theories and beliefs" Roth and Lawless (1999, pp. 369-373) point out that practitioners such as Gooding and Pickering "provide clear indications that language emergence is deeply caught up in material practice". The DM scientists narrated how technical skill and theoretical researching through material activities such as model building facilitated exploration and understanding of the scientific hypothesis. Ochs et al. (1994, p. 151) also explore the role of linguistic and material representations and describe how physicists "transport themselves by means of talk and gesture into constructed visual representations". The DM scientists reconstructed themselves as creative and they also provided a commentary on what it was like to accomplish these tasks. The importance of relationships to the successful outcome and to the personal meaning of success echoed Knorr-Cetina's (cited in Pickering 1992, p. 12) observations about the "making of social actors and relations alongside, and in mutual accommodation to the making of the material world of facts, phenomena and instruments".

Dunbar's studies on the "distributed process of science" (cited in Oatley 1996, p. 137) were directly relevant to the scientists' interviews. He refers to how conceptual change was engendered through regular team meetings where informed scientists from related backgrounds presented on a topic, which then allowed the concept to be reframed and progress made. Analysis of the DM scientists' narratives showed how they spoke of the sharing of ideas, but also the way an idea could become apparent to one part of the collaboration in the light of the specific work they had previously been doing. In this way collaborations were not all making progress uniformly, although it was the sum of the contributions that defined the successful team.

Oatley (1996, p.123) further refines Bruner's distinction between the narrative mode "for thinking about human action and a paradigmatic mode for thinking about mechanisms and natural science" in his assertion that the narrative mode is essential to scientific explanation. It provides a way for the scientists to communicate scientific principles to those outside the scientific community. It is characterised by a lack of emotional engagement as discussed earlier and instead uses inferences and reasoning. However it belongs to the social world in the sense that inference is a generalisable concept, beyond the scientific community, and science needs previous examples, in the form generally of written hypotheses, in order to confirm or refute an idea and make progress.

The way this is validated is by social engagement in structured arenas of debate and according to Oatley (1996, p. 137) it is in these arenas that narrative again becomes emotional. This was true of the DM interviews where the emotional language of the scientists and the colour and vividness of their imagery related to reconstructing the dynamics of competition and collaboration around the time of gene isolation. The significance of social areas around these arenas of engagement such as conferences became apparent as the scientists discussed how work was done outside the official presentations and papers in working out who to trust and what information was valid. It was also pertinent to the narratives that described how the gene discovery was validated, and anticipation resolved, by mediation within the scientific culture regarding the official publications and acknowledgement of the successful collaboration. The way that attribution of success "can be seen as a textual process; ...a way of telling a story" (Myers 1990, p. 102) was referred to by Prof who observed that it was difficult to know the exact sequence of events surrounding the discovery.

The contingencies apparent in the scientific narratives were not reflected in the official publications and supported Harre's (1990, p. 83) observations that the process of research bears little resemblance to its final presentation. The reporting of accurate scientific information was separated from the temporal unfolding of the events as they occurred in the laboratory and the resulting narrative formed part of the narrative of science itself, rather than an episode of scientific practice. This complicating of scientific discourse by a "narrative of objectivity" was described by

Latour and Woolgar (cited in Harre 1990, p. 99) as "deindexicalization", whereby scientific facts attained independence from the culture within which they were produced. The use of the passive voice and conventions removing human agency privilege the "public face of a scientific discipline" (Hyland 1997, p. 21) and reinforce science as infallible.

The DM scientists were aware of these conventions and their interview accounts gave insight into the processes that were not apparent in the published work. Their accounts moved between awareness of "The interdependence between the communication system, the reward system, disciplinary prestige and the validation of knowledge" (Hyland 1997, p. 26) and insight into how this was accomplished in practice. Prof's account in particular showed awareness of the way these features of academic cultures "can mitigate against the more public tenets of science that emphasise cooperation, impartiality and the free exchange of information" (Hyland 1997, p. 29).

Toit (2003) asserts that there are close links between narrative and sense making as narrative involves retrospective accounts that stress plausibility, credibility and coherence. Riessman (cited in Franzosi 1998, p. 546) stresses the importance of preserving the structure of narratives in order to "respect respondents' ways of constructing meaning and analyse how it is accomplished". The scientific and family interviews utilised narrative as a way of communicating experiences that are not usually accessible, either because of a lack of culturally available repertoires within which to contextualise these experiences, or a lack of agency in appropriating an audience to listen to them.

In their evaluation of the career of the DM gene discovery Batchelor et al. (1996, p. 248) stated the need for research that "extends...and takes the processes of discovery into the various domains of their production and reproduction". The retrospective accounts of the DM scientists and the family interview accounts examine DM gene discovery from the narrative points of view of the two groups of people most closely associated with its impact. In doing so the emotional significance of DM on everyday life and everyday work is revealed and additional perspectives to the medical classification and scientific definition attained.

Emotivation: Motive and Emotion

Through the collaborative experience I saw how science could function and move quickly which made it fun. This was an important experience for shaping how I supervise students and select projects so that they have the potential to deliver "big" results. Making good quality friendships and believing you can live the dream even with limited resources have both been very important to me.

Bill PhD student

The motivation to achieve scientific progress has been recognised in sociological analysis of paradigmatic scientific arenas such as human genetics and has been analysed in such frameworks as power, networks and discourse (Latour and Woolgar 1979; Collins 1983; Gilbert and Mulkay 1984; Latour 1987; Pickering 1992; Ochs 1994; Franklin 1995; Batchelor et al. 1996; Atkinson et al. 1997). The expression of personal motivation and emotion offering insight into complex feelings and dynamics is less documented. Narrative analysis is significant as a method and as a way of formulating and expressing this meaning.

The DM research environment was unique in terms of the timing of the discovery in the field of clinical genetics and the combination of clinical, scientific and familial expertise at the Institute, but it was the additional ingredient of emotion that infused the endeavour and its subsequent reconstruction with meaning. In their retrospective accounts of the impact of the discovery of the DM gene the scientific team reconstructed dynamics, relationships and emotions in a discourse that gave additional insight into the culture of the research scientist.

The distinction between "big" and "small" science based on the contrast between routinised, large scale and well- resourced enterprises and a smaller team approach comprising "a more heterogeneous collection of scientists and objectives, largely organised and scrutinised from within" (Yearley 1988; Collins 2003; Bartlett 2008, p. 54) shares some common features with the DM gene isolation as a small science project. Through their narrative accounts the scientists portrayed the DM team as small in terms of resource and profile. However they narrated a focus on outcome and a key element that outweighed the resources, profile or size of the team. This was the "big idea" and the DM team expressed pleasure in David and Goliath tales where this intellectual resource was a defining feature of their success. In their

reconstructions of themselves as creative, lateral thinkers and the environment they worked in as flexible and resistant to defined management styles, the culture within which a good idea could flourish become more visible. Bartlett (2008, p. 75) notes how "Demands for creativity, and the fact that the expertise of science, with its emphasis on novelty and progress prevents codification of work processes, have shaped the organisation of work in small science". The DM project, with the privileging of intellectual over material resources, resistance to definition of management style, and sharing of cross-disciplinary resources demonstrated in addition that motivation engendered by pride in personal resources was exemplified in the shape of the "big idea".

In his discussion of the characteristics of the work associated with the Human Genome Project as so-called "big science" Bartlett (2008, p. 55) observed how "The highest ranking member of the workshop or laboratory delegates the dirty, the laborious, and the tiresome tasks – the tasks of low esteem – to more junior members". Narrative analysis of the accounts of the DM team facilitated insight into the observation of such work practices and procedures through interrogating their personal meaning. Kay, the DM technician, repeatedly gave accounts indicating the apparently mundane nature of her work but imbued these tasks with pride and personal meaning. This was accorded significance through her recounting of how her work eventually received public recognition and how this was for her "the star". Through narrative analysis it is not only the nature of the task that becomes apparent but also its relevance to the actor. Through contrastive rhetoric Kay succeeded in illuminating dynamics of motivation within the lab as she contrasted her work with the priorities of others who were motivated by success beyond the everyday routine of the laboratory. Scientific success, as demonstrated through the traces in the DM medical records, was iterative, incremental and built on everyday work. Narrative analysis showed how the personal meaning of work differed between individuals but how it was the combination of these different practices that enabled success. The nature of the individual work tasks was less important than the recognition by others of successful and committed accomplishment of those tasks.

I use "Emotivation" in this thesis to describe the investment of self and of personal skills that characterised the narratives of the DM gene discovery scientists. This

added an additional emotional register to Gilbert and Mulkay's contingent repertoire, and was used in parallel to the empiricist repertoire representing the institutional or public face of science. Analysis of the DM scientists' interviews offered insight into the investment of emotion and motivation in the work of the laboratory. In doing so analysis moved beyond the identification of specific dynamics and discourses in the representation of scientific work, to awareness of how these dynamics are interpreted and their personal meaning for the actors themselves.

The importance of emotional engagement with work was personal but also a feature of the DM workplace. As the analysis showed this emotional engagement varied in personal meaning between the scientists but reference to relationships and dynamics were common across all the accounts. The laboratory, and for Prof this extended to the clinic and to family homes, was invested with significance through narratives highlighting what it was like to work in a place and on a project which could not be repeated. The observation that "...the recruitment of sentiment did produce real effects in the way that research participants worked" (Bartlett 2008, p. 185) was further refined by narrative analysis to reveal what was meaningful to the different members of the team. Through their accounts the DM team narrated the legacy of the gene discovery for them personally and the workplace as the site of memories, past valued reconstructions of self, and a unique scientific endeavour.

In their classic analysis of how cultures and phenomena are constituted by discursive practice Gilbert and Mulkay (1984) contrasted the empiricist repertoire of science as infallible, with the contingent repertoire which provided "the resources whereby scientists are able to account for the discrepancy between their own, empirically rendered versions of reality and those of their colleagues who seem to see the world differently" (Toolan 1998; Locke 1999, p. 122). In Potter and Wetherell's (1987, p. 208) development of this work on discourse they defined interpretive repertoires as "recurrently used systems of terms used for characterizing and evaluating actions, events and other phenomena". Edley (2001, p. 202) makes further conceptual and methodological distinctions between discourse and repertoire, where discourse is associated with power and repertoires are "smaller and more fragmented...and place more emphasis upon human agency within the flexible deployment of language".

Analysis of the DM interviews showed use of both of the empiricist and contingent repertoires and awareness within the scientific team of their context dependent use. This was made explicit through metaphorical references to two different worlds of the laboratory and the outside world. The contingent repertoire used by the DM scientists functioned in moral tales and contrastive rhetoric to illustrate good and bad scientific practice. It was additionally characterised by emotion, reference to relationships and personal meaning. This additional emotional register counterbalanced the recognition of public success, signified in the published scientific articles, with more private reflection of the personal impact of DM gene discovery. The emotional register functioned within the scientific narratives to give insight into the everyday work that resulted in scientific success. The accounts explored the motivations of the individual team members and how they influenced the outcome of the research. The emotional register extended awareness of repertoires of public and private discourse to attribute personal meaning to the relationships and emotions that are part of everyday work within the laboratory. Through attribution of meaning, given equal significance to the empiricist repertoire in the accounts, the use of the contingent repertoire is extended beyond a way of accounting for human fallibility to validating this everyday world of practices and relationships. It is not just that the empiricist repertoire visible in the published accounts of discovery presents the "facts" and the contingent repertoire describes a shifting world where things could have been otherwise, but the emotional register additionally describes a culture where, through human interaction, tasks are accomplished and accorded meaning. The DM scientists constructed narratives where the facts they have contributed to scientific knowledge are beyond dispute but where, through attribution of emotions and personal meaning, the work of gene discovery is given a human face and becomes visible as everyday work.

In their introduction to "Pandora's Box" Gilbert and Mulkay (1984, p. 2) assert that the "authorial voice of the sociologist" can dominate sociological analyses and that only actors appear who endorse this authorial point of view. The DM scientists reconstructed multiple memories and accounts of DM gene discovery. Narrative analysis allowed the context of these accounts to remain intact and highlighted the way in which these stories were given meaning through a vocabulary of emotion. The authenticity of the DM accounts therefore resided not with an attempt to fit a

single unifying theme or story of gene discovery, but with the speech acts of the actors themselves as they narrated an episode of scientific success and the grounding of this work in emotions, motivation and relationships.

Anticipation

> *Initially she thought it was arthritis as she is fifty seven, but when her brother was recently diagnosed with MD she was given a leaflet by him suggesting that she seeks genetic counselling.*
> *He [brother] began walking strangely and slowly and shuffling at least ten years ago. He has speech problems. He is very unsteady and can't go alone – is taken everywhere by his mother. He is now registered disabled. Mrs...is now very concerned that she may have MD due to her aching legs. She is particularly concerned because in the leaflet it mentions testing. Her daughter is currently 20 weeks pregnant with her 1st child.*
> *We discussed the importance of her daughter and her Obstetricians being aware of the family history of MD. Although her daughter knows that her uncle has the condition, she has not realised the implications for herself. Mrs...is reluctant to inform her daughter of any risks as she doesn't want to alarm her at this stage in pregnancy. I explained that I felt it was important for her and her obstetrician to be aware, but that I would discuss this further with Dr [geneticist] and telephone her to let her know what his advice is...I asked Mrs...to bring photographs to clinic...Mrs...was aware that her brother had a DNA test which showed he had MD but did not know what this meant...Mrs...knew that she had a risk of MD but did not know what this was...Mrs...says that she knows little about MD but knows that there is no cure and no treatment.*
>
> *Pre-clinic report from genetic nurse specialist in DM medical record 2005*

The discovery of the DM gene provided molecular evidence for the phenomenon of anticipation, whereby DM may manifest earlier and with increased severity through successive generations of a family. The potential impact of DM diagnosis on three generations is evident in the above extract. The anticipation of a new grandchild becomes part of a discourse about risk where previous and future generations are evaluated in the context of an inherited medical condition.

The decisions made by family members, the dilemmas described and the concerns shared did "not imply resignation in the face of an implacable biological destiny" (Novas and Rose 2000 p. 506) although a key theme was the difficulty retaining agency. Family members contrasted negative experiences of being defined by their DM diagnosis, with an ideal scenario locating DM within a discourse of everyday life, and being able to access help, advice and resources as necessary in order to maintain valued roles. The DM medical records documented the stress of living with

an uncertain diagnosis in the years preceding gene discovery. Incorporation of a diagnostic blood test shortly after DM gene isolation enabled a more rapid resolution of diagnostic uncertainty. The DM notes also document the self –referral of extended family members to their GP following diagnosis in a relative and the awareness of the gene test itself. Both the DM interviews and DM records highlighted the complexity of family dynamics within which the DM discourse is located. Although the test is available for individuals genetic counselling continues to take place within a wider framework of family relationships and these dynamics remain an integral part of the work of the clinic.

The adoption of a moral stance that "if you start from a position where genetic technologies are bad, then any assessment of these technologies is likely to conclude that they are indeed bad" (Hedgecoe 2001, p. 307) risks alienating people who have made decisions to utilise genetic technologies on the grounds of family history, and also places a genetic definition onto people who have chosen to have children despite a risk of genetic conditions. The fact that many people directly implicated through family history chose not to attend any form of genetic counselling or medical management when diagnosed shows that individuals have the capacity to make their own decisions about locating themselves within a genetic discourse. Whilst the possibility of truly non-directive counselling has been questioned the possibility of families own decision-making should also be recognised otherwise their experiences of having a genetic condition may lead to them being perceived as victims (Clarke 1991).

DM research and service began at the Institute in the early 1970's before the use of terms such as "geneticization" defined as "reducing people to their DNA codes" (Hedgecoe 2003, p. 51; Lippman 1992; Basen et al. 1993). The way that families spoke of their experiences of DM, some of which were shaped by decades of living with DM, did not reflect this reductive aspect. Interpretation of family interviews gave insight into a small group of people with an uncommon genetic condition who are linked with medical genetics through personal circumstance. Rose and Novas (2000, p. 485) speak of the "rise of the new molecular genetics" and its potential creation of an at-risk society. The DM family interviews illustrate a world of clinical

practice and family concerns grounded in everyday life, with reproductive risk predominating at key junctures of personal biography.

There were many different interpretations of what DM meant within a family but there was a consensus as to the implications for reproductive decision-making in the next generation. Men and women discussed this equally as did those who had DM and their unaffected partners. The emphasis on dealing with everyday life changed to discussions of educating a child about DM on reaching reproductive age. For the men interviewed who had no children this was an active choice to ensure that DM was not passed to the next generation. Where DM was not considered an issue in the present it was discussed in as a factor in future relationships. In this way DM was part of everyday life in the present, and as a genetic condition in relation to the future. The present was also characterised by the continual surveillance and monitoring of other family members to assess whether they had DM, and by a search for clues to a personal future that could be gained from observing how DM manifested in relatives.

Family was invoked through descriptions of emotional ties and relationships. Family was also constructed as offering alternative ways of coping with DM. Rather than being defined as a family with a genetic condition several interviews focused on accounts of how their family had a common attitude or strategy such as "we have a laugh about it". This offered another interpretation of how family is constructed and the personal meaning individuals associate with their family.

The contemporary understanding of family as composed of blood relatives and in-laws or relatives through marriage introduced a more complex interpretation of family (Strathern 1992a, 1992b). Narrative accounts of relationships with in-laws were constructed around DM as a site of delineation between families. DM was narrated as a factor influencing whether couples should marry, have children or carry on with pregnancy where DM has been diagnosed. This did not preclude the maintenance of relationships, such as with grandchildren diagnosed with DM, but the articulation of presumed responsibility for DM within families was a common theme in the interviews.

The challenges in genetic counselling in DM could be traced through the letters to patients after clinic consultation. Early letters indicated relative risk and degree of diagnostic certainty based on clinical expertise. Letters after gene discovery varied according to the clinical geneticist but the lack of absolute certainty was indicated together with the fact that "as we learn more about each condition so it may alter over time." The "slightly confusing thing about DM is the nature of the gene repeat" and the fact that it may increase "and for this increase in size to be quite a big jump" highlights the uncertainty that is still associated not just with DM but with communication of a concept that is itself the subject of continuing speculation. (Hedgecoe 1999; Shea 2001).

The researching, documenting and management of uncertainty characterised much of the work of the DM clinic. This included the reductive scientific application of probability, for example by linkage analysis, before the DM gene was identified. The management of DM by the geneticist through monitoring of systems such as the heart utilised a protocol that could be followed at every clinic visit, allowing a medical picture of the condition to emerge over time. The potentially fatal consequences of altered heart function could be addressed and some control over the uncertain prognosis exerted. Uncertainty as a part of the clinical encounter has been documented, as has the way in which clinicians are trained to deal with certainty (Atkinson 1984; Fox 1989). The uncertainty in the DM clinic was notable for the way in which consultations before the advent of the gene test were predicated on a degree of diagnostic uncertainty, whilst consultations post-gene discovery entail negotiation of prognostic uncertainty. The gene test was a landmark development but the medical records indicate the smaller incremental steps that were continually taken in the application of new knowledge to clarify uncertainty.

The aspects of DM management documented in the DM medical records show the extent to which the clinical geneticist has been involved with the implications of the diagnosis in everyday life. This has varied according to the visibility and remit of other personnel in the records. The involvement of clinicians in areas beyond the immediately medical has been classified as part of medicalisation in that it extends the power of the legitimacy of the clinician beyond the medical and into the social domain (Conrad 1992, p. 216). However the delegating of these aspects when

possible indicate that they were considered an essential part of DM monitoring, as evidenced by their place on the DM protocol, but not necessarily intrinsic to the work of the clinical geneticist.

Silverman (1993, p. 132) advises the researcher of documents to "...focus on how such files reveal the practical decision-making of employees in the context of the constraints and contingencies of their work". The DM medical records document continual adaptations in the face of developing knowledge and technology. This is particularly apparent in the issue of risk and reproduction. In medicine "Competence is at the heart of professional practice, its legitimisation, certification and everyday evaluation" (Atkinson 2004, p. 13). Continuous reappraisal was necessary to fulfil these criteria in the discipline of clinical genetics that was emerging at the beginning of the DM research and which has subsequently been integrated into routine medical practice. This is represented numerically and linguistically in the medical records in the evaluation of risk prior to gene discovery. As technology advanced new information became available which in some cases altered the risk percentage individuals had been given and impacted on their reproductive choices. The gene discovery was followed by the development and implementation of DM gene testing. The way that everyday work is composed of "interpretive practices" (Collins 1983, p. 90) that become visible only when they go wrong could be seen in the occasional references in the DM medical records to problems in the laboratory affecting gene test results. This highlighted the contribution of the laboratory to the smooth functioning of the clinical encounter (Atkinson et al. 1997). The negotiation between clinical judgement and molecular proof also became apparent in rare instances when a test result appeared to exclude, rather than confirm a clinical diagnosis.

According to Escobar et al. (1994, p. 211) "any technology represents a cultural invention, in the sense that it brings forth a world; it emerges out of particular cultural conditions and in turn helps to create new ones". The context of the isolation of the DM gene was defined by the ascendance of the genetic paradigm and the gene test offered resolution to a clinical and familial issue. However Franklin's (1995, p. 178) critique of terms such as "technoscapes" highlighted the danger that "a tendency towards hype attends closely on the heels of wonderment". The family interviews and analysis of the DM records indicate that the gene test changed the

climate of uncertainty surrounding diagnosis in many cases, but many of the difficulties associated with DM remain. Improvements in medical care, such as cardiac screening and drug treatment for excessive sleepiness, are associated with greater knowledge of the condition, but issues such as access to equipment and services remain mediated by agencies where lack of knowledge about DM and pressure of resources affect provision.

Sociological insight into the varied discourse of people with genetic conditions and the very different interpretations and decisions they make illustrate the socially situated way in which medical and scientific information is assimilated. Franklin (1995) distinguishes between information given in the clinical setting, and moral and cultural knowledge. She discusses how "common diagnoses such as Down's syndrome intersect widespread cultural knowledge and established medical certainty" (1995, p. 175). The DM family interviews highlighted shared themes, such as uncertainty or worry for the next generation, but this did not mean that the same actions were taken by all of the families as the issues are interpreted in the context of individual situations.

The advent of the new genetics resulted in warnings that it has "medicalized family and kinship creating profound ethical and practical dilemmas for both the individual and for medicine as a whole" (Finkler et al. 2003, p403). Prof introduced the context of his work as the early realisation that families with DM were living in dire circumstances and that nothing was being offered to help them. This concern may reflect a historical approach to the role of the research clinician but it illustrates the effects of conditions such as DM on the social circumstances of some families before the new genetics. Valid sociological concern of the risk of stigma through genetic labelling may not communicate the extent to which stigma or discrimination may occur, regardless of whether DM is classified as a genetic or neurological condition (Wertz 1992).

Epiphany

He [church minister] treats him [Leo] normal but makes allowance [for DM]. He called him out to do what he knew Leo could do...but he provided a chair for him. I thought what? I was just overwhelmed, I mean really overwhelmed. For somebody quite young to show such maturity and to be self-controlled to treat somebody exactly as normal and then accommodating his illness and I thought that's the secret.

Sarah, Leo's wife

The epistemological position of this research reflected work by practitioners such as Kerr and Kleinman in their advocacy that the meaning and the experience of participants be represented (2004; 1998). The grounding of research undertaken by health professionals and sociologists "in an understanding of everyday family practices that is sensitive to their complexities" counteracts the danger of "idealized and over generalized views" (Featherstone et al. 2006, p. 149). The family members narrated complex family dynamics, the variability of the impact of DM, and the importance of maintaining valued roles within the family and wider community, and in doing so made visible the complexity of the DM phenotype and the context and environment within which it is being expressed.

Halliday (cited in Hyland 1997, p. 29) drew attention to how "By their everyday acts of meaning, people act out the social structure, affirming their own statuses and roles, and establishing and transmitting the shared systems of value and knowledge." Lawton (2003, p. 39) calls for medical sociologists to be"more open-minded to the use of novel and seemingly unconventional theoretical and methodological approaches". In this thesis narrative offered a method and a way of representing different perspectives. In their narrative accounts the DM scientists reconstructed the emotional and interpersonal work that was part of the everyday work of scientific discovery. The narratives challenge the stereotypical image of the scientists as detached and impersonal, and move the reference point of DM in families from being a patient to the achievement and maintenance of roles of everyday life.

The impact of the accounts lay in their emotional resonance and the agency of the key actors in narrating the impact of gene discovery, as it was meaningful to them. The resulting emphasis on the personal meaning of relationships and dynamics in everyday life and everyday work highlighted that "the epiphanies that move us are not the marvels of life, but its very ordinariness" (Bruner 1996, p. 98).

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Appendix one

a. Interview schedule for gene discovery team submitted as part of Cardiff University School of Social Sciences ethics application.

At the beginning of each interview there will be a short introduction to explain again the purpose of the interview and the form it will take.

Examples of questions:

In your career working on DM what were the major highlights for you?

How did you become involved in research for this condition?

Are you still working in this area?

What changes have you seen since the gene was discovered?

What implications have these had for you in your working life?

What effect have they had on your everyday work with patients and families?

Did you anticipate any of the ethical issues, such as testing or insurance, before the genes were discovered?

Do you think that gene discovery has changed the questions which families ask you when they come to clinic?

Has your work practice (lab work/genetic counselling) changed over the past 20 years?

Has the genetic counselling experience changed for families?

The wording of the questions will vary depending on whether the respondent is a scientist or clinician. The opportunity to move onto other questions and areas of enquiry will be left open but the interview will aim to cover the above questions, not necessarily in order. The first question is designed to open up the area of enquiry and then allow for questions on more specific issues such as possible changes in clinical practice. One key aim of the interview is look in detail at if changes in practice were brought about, and if so is there a route of change which can be traced. Another aim is to give the key gene researchers the opportunity to talk about their experiences of a time when events moved very fast and which they may not have reflected on in depth subsequently, although some are still actively involved with the particular gene.

b. Interview schedule for families with DM submitted with NHS COREC application for ethical approval.
Interview Topic Guide for Participants

Version 1.0, 27.10.2006. For LREC approval.

The interviews will be informed by the pilot study of medical records in the myotonic dystrophy archive. The following are expected to be areas of interest but additional topics may arise. The interviews will be semi-structured in order to allow maximum input from the participants. The researcher will use an aide-memoire as a guide during the interviews.

Themes

- Initial introduction to myotonic dystrophy
- Previous awareness of myotonic dystrophy and of heredity conditions
- Dealing with changing knowledge about an ongoing condition
- The effects of scientific discovery on understanding of the condition
- The effects of scientific discovery on the practical issues of living with myotonic dystrophy
- Involvement with research
- Ways in which interaction with the genetic clinic has changed

Communication about a genetic condition as knowledge changes and scientific advances are made.

Appendix two.

Introductory letter, Participant information sheet and Consent form approved by NHS COREC application. Thank you letter as part of the research protocol.

a.

The impact of gene discovery on patients, doctors and clinics

Letter of invitation. Version 1.0. 27.10.2006. On University of Wales headed paper. For LREC approval.

Dear (name),

I am writing to you as you attend muscle clinic at the University Hospital of Wales and agreed that you were happy to be contacted about any research projects related to myotonic dystrophy. This is on the understanding that you are under no obligation to take part.

I enclose an information sheet about a project which may be of interest to you. The project relates to ways in which the discovery of the gene for myotonic dystrophy may have changed the genetic service, or your own personal experience of myotonic dystrophy. The project also involves looking through the clinical records held at the Institute of Medical Genetics, University Hospital of Wales. I have discussed the project with..., muscle clinic consultant, who is the head of the muscle service.

If you would be willing to take part, then do read the attached information sheet.

- Part 1 tells you the purpose of this study and what will happen to you if you take part.
- Part 2 gives you more detailed information about the conduct of the study.

Please take your time to consider whether or not you would like to take part. I will contact you one week after sending this letter to ask for your decision. If you would like to contact me to discuss the project before then please do not hesitate to do so. Please feel assured that there is no need to take part if you do not wish to.

Yours sincerely,

Cathy Sampson

b.

The impact of gene discovery on patients, doctors and clinics

Research Participant Information Sheet – Part 1.
Version 1.0. 27.10.2006, on Cardiff University headed paper. For LREC approval.

What is this project about?

The project aims to look at how the discovery of the gene for myotonic dystrophy affected patients, doctors and the genetic clinic. The Institute of Medical Genetics at the University Hospital of Wales was involved in the research leading to the discovery of the gene but also has many years of experience of running the muscle clinic. The co-operation of families and doctors has been vital and this study wishes to look back at how the experience of being diagnosed with and living with myotonic dystrophy has changed over this time. The project is funded by the ESRC as a PhD studentship for the researcher, Cathy Sampson.

Why is this being studied?

There have been other studies looking at the gene discovery itself but none looking at the everyday way in which gene discovery affected everyone involved in the clinic.

Why have I been chosen?

You are being invited as you have had a diagnosis of myotonic dystrophy and attend Muscle clinic at the University Hospital of Wales. We would like to interview approximately twenty people for this study.

Do I *have* to take part?

NO. And if you join in but then change your mind, you can withdraw at any stage. Whether you do or do not take part will make no difference to the medical care provided to you and your family in any way.

If I take part what would that involve?

Cathy Sampson, the researcher for the project, will contact you one week after you have received this information sheet. You can let her know whether or not you would like to take part and will have the opportunity to ask any further questions. If you would like to take part Cathy Sampson will arrange to meet you for an informal interview lasting approximately one hour. This could be in your home or elsewhere – as you wish. Before starting the interview you will be asked to sign a consent form and asked for permission to look at your medical notes and to have an informal conversation with you. You can withdraw from the study at any time and your data can be destroyed immediately.

Would anyone else in my family be involved?

The researcher will only contact family members who have requested information on research projects from ... at the Cardiff muscle clinic, or who have given signed consent to be part of the Myotonic dystrophy research register. Should one of your family wish to take part they should request this through the Cardiff muscle clinic.

How long do I have to think this over?

You will be contacted approximately one week after receiving this information letter. However you may tell the researcher, Cathy Sampson that you would like more time to consider the project.

What if there is a problem?

Any complaint about the way you have been dealt with during the study or any possible harm you might suffer will be addressed. The detailed information on this is given in Part 2.

Will my taking part in the study be kept confidential?

Yes. All the information about your participation in this study will be kept confidential. The details are included in Part 2.

If I want to talk to someone about the project, whom can I contact?

If you would like to talk to someone about the project, before you take part or afterwards, then you could contact ...in the Institute of Medical Genetics at ...telephone .... You could also contact the researcher, Cathy Sampson by telephone ...or by email... No confidential information can be discussed via email and therefore email can only be used for practical arrangements.

The impact of gene discovery on patients, doctors and their clinics.

Research Participant Information Sheet – Part 2.
Version 1.0, 27.10.2006, on Cardiff University headed paper. For LREC approval.

What if there is a problem?

If you have a concern about any aspect of this study, you should ask to speak with the researcher, Cathy Sampson, who will do her best to answer your questions. You can contact her by telephone on ...or by email at... If you remain unhappy you can contact ...in the Institute of Medical Genetics ...telephone ...

Will my taking part in this study be kept confidential?

Our procedures for handling, processing, storage and destruction of data are compliant with the Data Protection Act 1998. With your permission the researcher will look at your medical notes and may then ask you if they can audio tape record a conversation with you. All the information about you which is stored on computers

will not have your name on it so you cannot be recognised from it. Audio tapes will be kept in a locked cabinet and will be destroyed after the completion of the study.

What will happen to the results of the research study?

At the end of the project we will produce a short summary of our overall findings which we will send to participants. The summary will also be available on request. The results may be published so that other people working in this area could access the information. It will not be possible to identify any participants from this summary or from any other publications.

Who has reviewed the study?

This study was given a favourable ethical opinion for conduct in the NHS by the Research Ethics Committee.

Thank you for considering taking part and taking time to read this sheet.

c.

The impact of gene discovery on patients, doctors and their clinics.

Consent Form Version 1.0, 27.10.2006 to be on Cardiff University headed paper for LREC approval

I have read the information sheet about this research project and have had a chance to discuss my questions with the researcher.

YES/NO

I understand that there is no need for me to take part and that I can withdraw from the project at any stage.

YES/NO

I understand that there is no benefit to me from taking part.

YES/NO

I am willing to take part in the project and agree to allow the researcher to look at my medical notes.

YES/NO

I am willing to take part in the project and agree to have my conversation with the researcher audio recorded.

YES/NO

When the project is reported I understand that it will be anonymised so that I could not be identified.

YES/NO

SIGNATURE: **Date:**

NAME: **Date of birth:**

ADDRESS:

.....

I confirm that I have explained the nature of this research project to (NAME), that they understand what this involves and that they are willing to participate.

SIGNATURE: **Date:**

RESEARCHER NAME:

d.
Thank you letter to families following interview (not part of LREC application)

Dear

Thank you for your time when I visited you. It was very helpful and interesting and I appreciate that some of the topics may have been difficult to discuss. Please do not hesitate to contact me if there is anything further you would like to talk about in relation to the interview.

With best wishes,

Cathy Sampson.
Research Student

Appendix three: Data collection sheet for DM archive

Section one: **Physical Layout**

File number
Stored
Access
Colour
Condition
Size
Part of family set
Chronology
Layout

Front cover

Referral

Date
Who
What
Format
Active
How many people referred

Characters

Who is in the notes?

Family

Professionals

Section two: **Letters**

How many:

Who from	Who to	Typed/ handwritten	When	Form of address	Reason

Section three: **Official reports and standard forms**

How many:

Who from	Who to	Date	Particular language	Reason

Section four: **Handwritten section**

Date of first entry
Date of last entry

When	Who	Why	Layout

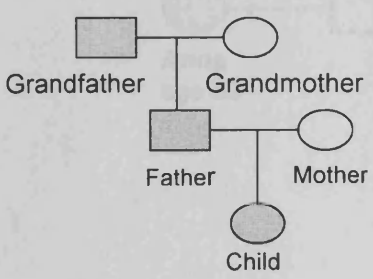
Section five: **Chronology**

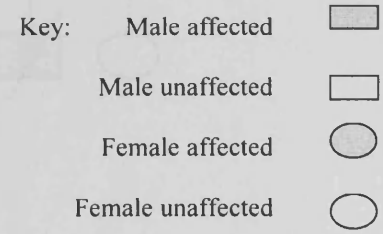
Section six: **Interesting language**

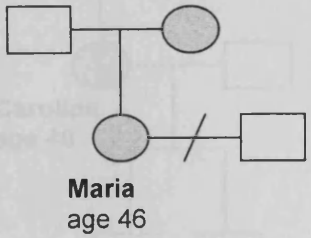
Who	to whom	When	Subject

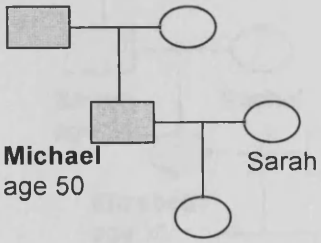
Appendix four: Family trees for DM family interviews

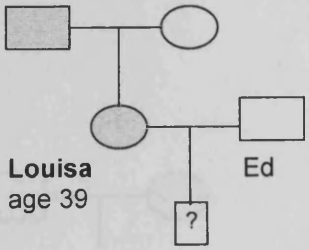
Legend: To protect confidentiality pseudonyms are used but family relations are represented correctly. Family members interviewed are in bold.

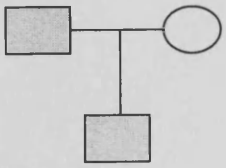


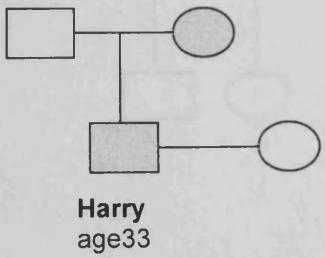


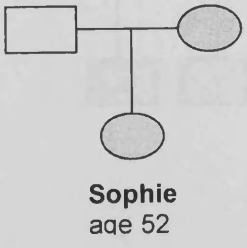


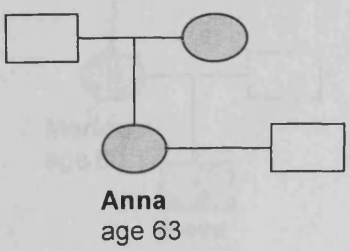


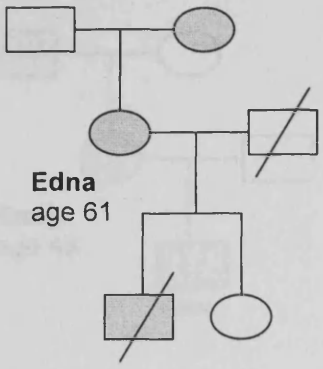


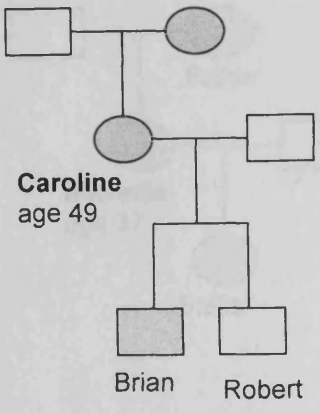







Anna
age 63


Edna
age 61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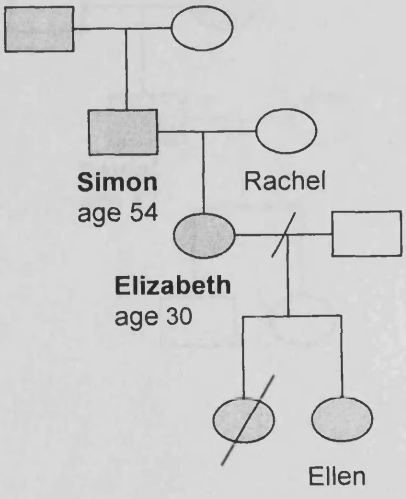
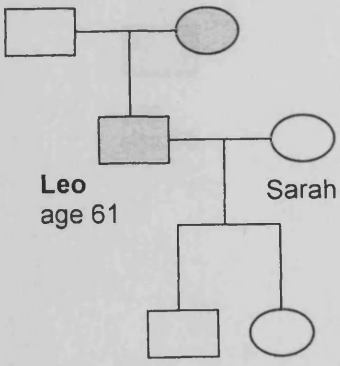
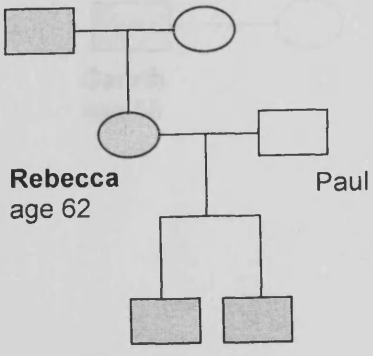
Brian
Robert


Simon
age 54
Rachel
Elizabeth
age 30
Ellen


Leo
age 61
Sarah


Rebecca
age 62
Paul