VOLUME I

RESEARCH COMPONENT

AN INVESTIGATION OF SOCIABILITY: DELINEATING A BEHAVIOURAL AND SOCIAL PHENOTYPE FOR MONOSOMY 1P36 DELETION SYNDROME

by

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ABSTRACT

Background: Research on Monosomy 1p36 deletion syndrome indicates there may be behavioural characteristics associated with the condition. However, there is no specific research on the social and behavioural phenotype of the disorder. The primary aim of this study is to delineate the behavioural phenotype for the condition, with a particular emphasis on the social phenotype by comparing individuals with Monosomy 1p36 to matched individuals with three other genetic syndromes (Angelman, Cri du Chat, and Cornelia de Lange) on measures of social behaviour and to observe social behaviour in experimental social presses.

Method: 90 participants were included in the comparison study, aged between eighteen months and forty five years. Twelve individuals aged between three years three months and thirteen years eleven months who had a confirmed diagnosis of Monosomy 1p36 deletion participated in the observation study. A number of behavioural measures were employed and individuals were observed interacting with a familiar and unfamiliar adult where adult engagement/attention was manipulated across five conditions. Video recordings of the observations were coded for social behaviours and skills.

Results: Results from the comparative study indicate impaired social communication, lowered mood and higher sociability with familiar adults are all notable characteristics for 1p36. In the social presses, individuals were more social under conditions of high attention/engagement with both familiar and unfamiliar people.

Conclusions: The study is the first to investigate social behaviour in 1p36 syndrome and as such the conclusions drawn are tentative. There is evidence that some characteristics may form part of a behavioural and social phenotype for the condition.

OVERVIEW

Volume I of the thesis is the research component and contains three papers. The first is a literature review of the definitions and measures employed for key aspects of sociability in individuals with intellectual disabilities prepared for submission to Research in Developmental Disabilities. The paper reviews the available definitions for four concepts related to sociability (social cognition, social competence, social skills and social behaviour) a concept which itself is poorly defined. By reviewing the definitions available in the wider social and cognitive psychology literature and comparing these to definitions provided in research with individuals with learning disabilities it is proposed that some of the concepts are poorly defined. The current article suggests possible working definitions which may be used as the impetus for future debate in the area. The clinical implications of having implicitly understood concepts rather than definable and measurable traits are considered. The review calls for researchers to provide definitions for the concepts being investigated and to consider the measures employed. The second paper presents an empirical study exploring the behavioural and social traits of a rare genetic syndrome, Monosomy 1p36 Deletion syndrome. The primary aim of this study is to delineate the behavioural phenotype for the condition, with a particular emphasis on the social phenotype by comparing individuals with Monosomy 1p36 to matched individuals with three other genetic syndromes (Angelman, Cri du Chat, and Cornelia de Lange) on measures of social behaviour and to observe social behaviour in experimental social presses. Results from the comparative study indicate impaired social communication, lowered mood and higher sociability with familiar adults are all notable characteristics for 1p36. In the social presses individuals were more social under conditions of high attention/engagement with both familiar and unfamiliar

people. The study is the first to investigate social behaviour in 1p36 syndrome and as such the conclusions drawn are tentative. However, it is suggested that there is evidence that some characteristics may form part of a behavioural and social phenotype for the condition. This paper represents the empirical research study of the thesis and is prepared for submission to the American Journal of Medical Genetics. The third paper is a public domain briefing document, which summarises the literature review and gives a brief overview of the empirical study. This is presented in appendix 2 of volume I. Instructions for authors and notes for submission to the journals are presented in appendix 3 of volume I.

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1. Literature Review

A Review of Defining and Measuring Sociability in Children with Intellectual Disabilities

ABSTRACT

There is a substantial body of research indicating that compromised social functioning for individual with intellectual disabilities can have far reaching implications for quality of life, community participation and well being. As the implications of such findings are so important for people with intellectual disabilities the research has grown at a fast pace. However, an inherent difficulty for research on social functioning is the lack of definitions for key concepts in the area. The current paper reviews the available definitions for four concepts related to sociability (social cognition, social competence, social skills and social behaviour) a concept which itself is poorly defined. By reviewing the definitions available in the wider social and cognitive psychology literature and comparing these to definitions provided in research with individuals with learning disabilities it is clear that some of the concepts are poorly defined. The current article suggests possible working definitions which may be used as the impetus for future debate in the area. The clinical implications of having implicitly understood concepts rather than definable and measurable traits are considered. The review calls for researchers to provide definitions for the concepts being investigated and to consider the measures employed.

INTRODUCTION

The importance of social functioning for individuals with intellectual disabilities has long been recognised through acknowledgement of its importance for an individual's quality of life, wellbeing and ability to participate in their community (Nota, Ferrari, Soresi, & Wehmeyer, 2007). Given the importance of the concept it is unsurprising that it has received so much research interest. However, a brief review of the literature reveals numerous different uses of many terms related to social functioning. The term sociability, for example, is often used as an umbrella heading encompassing numerous aspects of social functioning and is often not defined. Similarly, the constructs which fall under or are related to the term of sociability (e.g. social cognition, social behaviour, social skills, social competence, social functioning) are often used interchangeably and without reference to a definition, therefore making comparisons across research difficult.

Poorly defined terminology makes it extremely difficult to evaluate and integrate research and construct models of the determinants of social functioning. The difficulty of interchangeable concepts with no standardised definition was highlighted in a recent review of psychometric methods used to test children's social skills (Matson & Wilkins, 2009) where no definition of social skills was provided and yet over 40 tests were found for the construct of 'social skill' with tests not always covering social skills alone. If terms cannot be defined, it raises the question of how research can be replicated and generalised. There is clearly a need therefore to define and differentiate the many concepts related to sociability and social functioning.

The Concept of Sociability

The term sociability has begun to be used more frequently in recent years to describe numerous facets of social interaction and functioning. However, its use as an umbrella term can be

problematic when trying to understand the focus of research. One dictionary definition for sociability is "the relative tendency or disposition to be sociable or associate with one's fellows" (Sociability definition, n.d; a) and another states two levels of definition "the act or an instance of being sociable" or "the quality, state, disposition, or inclination of being sociable" (Sociability definition, n.d; b.). Neither of these definitions provides terms which could be operationalised for objective measurement, suggesting that the term 'sociability' needs further refinement in order to provide researchers with a clearly delineated concept to investigate. However, the term is used in research and one way that it is employed is as an umbrella term to cover various concepts such as social cognition, social behaviour and social skills. For this solution to be useful for research the concepts themselves need to be well defined and standardised across different studies.

There exists such a vast literature on the various aspects of sociability (taken as an umbrella term) in individuals with intellectual disabilities that an introduction to the area poses problems with given how poorly concepts have been defined. Research does show important relationships between different aspects of sociability which impact upon an individual's life. However, this research often does not state the meaning of the concepts being investigating; leaving the reader to infer what is meant by certain terms (e.g. social skills, social behaviour). The importance of understanding the impact of problems with 'sociability' for individuals with intellectual disabilities can be seen with a brief introduction to the area Again however, concepts are rarely defined.

Sociability in Intellectual Disabilities

In recent years there has been an increase in attention from researchers regarding the social skills (typically deficits in social behaviours such as eye contact problems, social interaction difficulties, lack of play behaviour etc.) of children, particularly children with intellectual disabilities, developmental disabilities and genetic syndromes. Whilst historically the literature has recognised an association between intellectual disabilities and social skills, the association has typically been

negative suggesting that intellectual disability leads to difficulties in social adaptation and functioning (Tregold, 1937).

Much of the research on sociability in children with intellectual disabilities would support such a claim and the importance of investigating sociability has long been recognised as a way to understand potential future difficulties children might encounter. Some of the findings indicate that deficits or impairments in social skills are related to numerous problems, including attention deficit hyperactivity disorder (Boo & Prins, 2007), social isolation and withdrawal (Chung et al, 2007; Matson & Boisjoli, 2007), aggressive and antisocial behaviour (Webster-Stratton, Reid & Hammon, 2001), and challenging behaviour (Fox, Keller, Grede & Bartosz, 2007). Of course, the research does not imply a cause and effect relationship between these variables and social skills, but would suggest that sociability is an important correlate.

Research on children with intellectual disabilities also suggests that social skills problems can be an indicator for other social problems such as social behaviour problems, deficits in prosocial skills and displayed aggression which can lead to poor peer relationships and social maladjustment (Bellanti & Bierman, 2000). The relationship between intellectual disabilities and peer relationships has also been highlighted in pre-school children who have been found to have compromised social skills leading to an inability to develop relationships (Guralnick, 1997).

It has been suggested that difficulties in social relationships for children with intellectual disabilities may be due to different or impoverished social interaction which in turn are due to delays in the development of interactive skills (Sheriden, Hungelmann & Maughan, 1999), or a lack of initiation of and maintenance of social interactions with peers (Kamps et al., 1992). Research has also suggested that specific difficulties exist which then impact on higher social functioning, for example, difficulty in appropriately interpreting social situations, including comprehending verbal and non-verbal social cues (Bruno, 1981; Markoski, 1983); problems focusing attention on social cues and instead paying attention to irrelevant information (Tur-

Kaspa & Bryan, 1994); and lower competence levels than typically developing children in taking others' perspectives and understanding others' intentions (Weiss, 1984; Wong & Wong, 1980). Furthermore, it has been suggested that children may show lower levels of socially interactive play with their peers and more socially isolated play, leading to further problems with peer relationships and from a young age (Kopp, Baker & Brown, 1992).

However, research has now begun to emerge on social skills and functioning which paints an altogether different picture for some children. Investigations into some genetic syndromes (Williams syndrome and Angelman syndrome in particular) have revealed some children to be excessively social (Jones et al., 2000; Oliver, Berg, Moss, Arron & Burbidge, 2009). It has also long been acknowledged that individuals with Down syndrome possess good social skills, are engaging and affectionate (Moore et al., 2002), show lower prevalence of aggression, attention seeking, untruthfulness and antisocial behaviour (Collacott et al., 1998) and have social communication skills and relationships comparable to typically developing control groups (Laws, & Bishop, 2004).

As the potential implications of problems with sociability are wide reaching, numerous intervention initiatives have been developed to try to ameliorate the impact. Indeed, much of the focus of research has been to establish clinical interventions to help improve social skills and social behaviour. The importance therefore of researching and understanding sociability in children with intellectual disabilities has been established. However, there is an inherent difficulty in researching and evaluating research in this area, namely, how to define the construct.

The impetus of the current review is therefore to provide an examination and understanding of how sociability (employed as an umbrella term) is researched and defined. The aim of the review is to define and differentiate the constructs used to assess sociability in children with intellectual disabilities (as found in the current literature); and to compare these to definitions of the constructs within social and cognitive psychology literature. This review will add to the working

definitions currently used to investigate sociability in children with intellectual disabilities to aid in future research.

Search Criteria

Psychinfo® and MedLine ® electronic databases were utilised to conduct a literature search using the search terms in Table 1.1.

Table 1.1: Terms employed in the literature search for studies examining sociability in individuals with intellectual disabilities or developmental disorders.

Search term	Variations
Sociability	Social behaviour & behaviour; prosocial behaviour & behaviour; social motivation; social competence; social skills; social interaction; social cognition; social perception; social reciprocity; social participation; social avoidance
Intellectual disability	Learning disabilities; intellectual disability; intellectual disabilities; intellectual impairment; developmental disorder; mental retardation; mental handicap.
Children	Child; children.

Papers were selected that had investigated aspects of sociability in individuals with intellectual disabilities or developmental disorders through the use of standardised or novel measurements. Papers were excluded if the participants were adults and/or no measurement of sociability/social traits had been used. Papers were also excluded if their primary purpose was the identification of children with autism, as numerous literature reviews already exist on the subject. However, papers describing methods for assessing social traits in children with pervasive developmental disorder or autism spectrum disorders were included. Papers were also excluded where the focus was on intervention programmes for social deficits. Only clinical and experimental studies and case studies from peer reviewed journals were included.

THE SOCIAL CONSTRUCTS OF SOCIABILITY

The literature search indicated that four main concepts or constructs are investigated in children with intellectual disabilities: Social Cognition, Social Competence, Social Skills and Social Behaviour. The accepted definitions from cognitive and/or social psychology of the constructs

investigated under the heading of sociability will now be reviewed and compared to the definitions provided in the intellectual disability research and ways in which they are measured as a way to begin to explore the consistency with which the concepts are investigated.

Social Cognition

The concept of social cognition is perhaps the easiest of the sociability constructs to begin to explore due to the vast amount of research on the topic. This of course does not mean that the concept will be easy to define and differentiate from other social constructs. It has been estimated that there are over 100 definitions of social cognition (Ostrom, 1994) and it can be assumed that many more have been developed in the 15 years since that estimate was made.

Perhaps the difficulty in defining the concept lies in its all encompassing nature; indeed Ostrom (1994) stated that the whole *Handbook of Social Cognition* (both volumes) should be taken as a definition for the construct (p. ix). Whilst numerous definitions exist, it does not appear that any are frequently cited as the accepted or definitive definition. Indeed, the majority of research appears to present the concept as implicitly understood.

One way of understanding social cognition has been to use a broad approach, such that individual aspects of social cognition (e.g. theory of mind, understanding another's perspective, social problem solving etc.) are not defined. For example, a recent definition is "social cognition is defined as the perception of others, the perception of self, and interpersonal knowledge" (Beer & Ochsner, 2006). A similarly vague definition has been provided by Frith and Blakemore (2006) "social cognition is defined as any process that involves other people". Whilst such definitions allow for a range of social cognitive processes to be investigated, they do not provide the research area with a clear definition of the construct under examination.

The main difficulty with defining social cognition as a unitary construct appears to be that it is not a single concept, but rather an approach or philosophy (Augoustinos, Walker & Donaghue,

2006, p. 16). In a review chapter on social cognition in individuals with intellectual disabilities, although a model of social information processing was referenced and adhered to, no definition of social cognition was provided (Leffert & Siperstein, 2002). However, seen as an approach or philosophy it no longer appears surprising that there is no agreed definition for social cognition. Rather, it makes sense that individual researchers outline the areas of social cognition in which they are interested and provide definitions for these.

Social Cognition and Children with Intellectual Disabilities

Given the above brief review of definitions employed in social and cognitive psychology to describe social cognition, it is anticipated that definitions for the concept will also be difficult to find in research with children with intellectual disabilities. Seven papers were identified that matched the search criteria and inclusion/exclusion criteria. Bauminger et al., (2005) defines the concept as:

> Social cognition includes the child's ability to spontaneously read and correctly interpret verbal and nonverbal social and emotional cues; the ability to recognize central and peripheral social and emotional information; the knowledge of different social behaviours and their consequences in diverse social tasks (e.g. how to initiate a conversation, how to negotiate needs, how to make group entry); and the ability to make an adequate attribution about the other person's mental state (i.e. "theory of mind" abilities or role taking abilities). (p. 45)

Six of the papers do not give a definition of social cognition per se, although some do provide definitions for the specific areas of social cognition they are researching. Leffert, Siperstein & Millikan (2000) define two areas of social cognition: social perception and social strategy generation. Social perception is defined as "an individual's ability to interpret or read relevant social messages from others. These messages, known as social cues, consist of verbal and nonverbal stimuli in the environment" (p. 531). They then describe what social cues are, making it easier for other researchers to examine the kind of social interactions and behaviours they are

referring to. Their definition for social strategy generation is "involves the ability to think of solutions for resolving social problems that are age-appropriate and that fit the immediate situation" (p. 532).

Whilst Cornish et al., (2005a) also do not give a definition of social cognition they do state that although they researched theory of mind and mental state understanding 'other components such as emotion and face recognition, eye gaze, social anxiety and perception are all important aspects' (p. 377), thus recognising that social cognition is not a unitary concept and has many facets.

Four of the papers (Bauminger et al., 2005; Gomez & Hazeldine, 1996; Leffert & Siperstein, 1996; Tur-Kaspa, 2004) primary aim was to investigate social information processing, and all cite the model proposed by Dodge (1986; Crick & Dodge, 1994). This model involves six steps: 1) encoding social cues from the environment; 2) forming a mental representation and interpretation of the cues; 3) searching for possible behavioural responses; 4) deciding on a response from those generated; 5) enacting the selected response; 6) enactment-including monitoring the effects of behaviour and regulating it. This model could be taken as a description of social information processing as applied by the above four authors.

The measures employed to assess social cognition can be seen in Table 1.2. The measures do relate to key areas of social cognition including social information processing, theory of mind, emotion recognition, social perception and social strategy generation. Five of the seven papers all use a similar methodology based around the social information processing model of Dodge (1986; Crick & Dodge, 1994) and adapted for use in individuals with intellectual disabilities (Tur-Kaspa & Bryan, 1994). Such a measure closely matches the constructs of the social information processing model and thus inherently appears to provide a definition for social information processing. Some measures of social behaviour are also included in some of the social cognition papers, which would appear to not match the construct under investigation. However, given that the fifth and sixth steps in Dodge's social information processing model both involve social

behaviour, it seems appropriate that social cognition should also be assessed in such a 'real-world' way.

Given the difficulty in defining social cognition in the wider literature, it is not surprising that researchers investigating social cognition in individuals with intellectual disabilities have also not established a widely accepted working definition of the concept. However, Bauminger et al., (2005) did provide a comprehensive working definition which coincides with the six steps put forward in the social information processing model (Dodge, 1986; Crick & Dodge, 1994) and the measures employed in the identified papers, and thus could be the starting point for a definition of social cognition in intellectual disabilities research.

Social Competence

As with many of the social terms, there is no universally accepted definition for social competence and the term has undergone various transformations and has evolved over time and throughout research. Early definitions focused on social behaviour, with cognitive elements being added later and, more recently, affective components have been seen as equally important (Topping, Bremner & Holmes, 2000).

Some definitions have been prescriptive, providing a list of dimensions thought to be important for social competence, such as problem solving, perspective taking and person perception (Sarason, 1981). Such an approach would make research consistent, if the list of domains could be agreed, as the presence or absence of each 'skill' could be measured. Other approaches have been far more flexible and offer little direction for researchers, such as the definition 'being well liked by peers' (Hubbard and Coie, 1994). It would be very difficult to operationalise such an open ended definition and to apply it consistently across different groups of children. Therefore, it would seem that a good working definition would need to strike a balance between the two approaches.

In recent years some definitions have tried to consolidate the two approaches and also acknowledge the impact of environment and culture on social competence. Topping et al., (2000) suggested "social competence is possessing and using the ability to integrate thinking, feeling and behavior to achieve social tasks and outcomes valued in the host context and culture" (p. 31). These authors go on to suggest that such a definition suggests a set of component skills are necessary for social competence and give some examples, but still this definition is difficult to standardise across research.

Perhaps the best way to begin the process of defining social competence is to see it as a broad term that encompasses other social concepts such as social skills and social behaviour, with an overarching theme of successful social outcomes that are pertinent to the context. However, until the concepts referred to by the construct of social competence are defined, there will remain a difficulty in defining the term.

Social Competence and Children with Intellectual Disabilities

As such difficulties exist in identifying a definition of social competence it will be interesting to see how the concept has been defined and measured in research with children with intellectual disabilities. Three papers were identified that purported to have investigated social competence in children with intellectual disability, using standardised or replicable measures.

None of the papers provide a definition of social competence although all acknowledge difficulties with the concept. Merrell and Popinga (1994) suggest that although there are different perspectives on what social competence is, there is a general agreement that the presence of adequate social competence allows individuals to have successful outcomes in social situations, develop positive relationships with peers and engage in social behaviours that have mutually reinforcing consequences (p. 40). Charman and Campbell (2002) discuss at length the potential relationship between theory of mind and 'everyday social competence and social behaviours', but fail to offer a definition of social competence. Whilst Rosner, Hodapp, Fidler, Sagun and Dykens

(2004) do not provide a definition of social competence either, they discuss the way in which social competence is assessed as typically being concerned with the number and quality of jobs, chores, friends, clubs and hobbies a person has; intimating that this is one way to measure social competence.

The measures used by the papers can be seen in Table 1.2. The majority of measures employed in the social competence studies measure aspects of social behaviour and an individual's ability to interact and 'get along with' others. Whilst these at first do not appear to be measures of social competence, when compared with the themes in the definitions of social competence above, it would appear that taken together some of the measures would satisfy a definition of social competence. For example, the Social Skills Rating System, the Frith, Happé and Siddons (1994) Sociability Scales and the Child Behavior Checklist all measure areas of social skills/behaviour; and the adaptive behavior domain of the scales of independent behaviour, the socialisation scale of the Vineland Adaptive Behavior Scale and the Child Behavior Checklist all assess social interaction and an individual's ability to interact with other people. Both of these themes have been highlighted as important concepts in social competence (Sarason, 1981, Hubbard and Coie, 1994).

Therefore, although none of the social competence papers provide working definitions of the concept, the measures they employ do imply what aspects of social competence they are evaluating. Further definition of social competence and differentiation from the concept of social behavior is needed.

Table 1.2: Measures employed by each study, with the construct reported to be under investigation, the measures and the constructs the measures report to assess.

Construct reported to be investigated by the paper	**Measures employed**	**What constructs are measured (based on the measure employed)?**
Social Competence (Merrell & Popinga, 1994)	• Social Skills Rating System (SSRS; Gresham & Elliott, 1990) • The adaptive behavior domain of the scales of independent behaviour (Bruininks et al., 1984)	• **Social behaviours-**2 subscales: social skills scale (cooperation, assertion, self-control and responsibility) and problem behaviour scale (externalising problems, internalising problems and hyperactivity). • 4 subscales: Motor skills; personal living skills; community living skills; social interaction and communications skills (**social interaction**, language comprehension, language expression)
Social Competence (Charman & Campbell, 2002)	• Socialisation scale of the Vineland Adaptive Behavior Scale (Sparrow, Bella & Cicchetti, 1984) • 2 16-item scales to assess 'active' and 'interactive' sociability (Frith et al., 1994)	• The socialization domain covers play and leisure time, interpersonal relationships, and various coping skills. Charman and Campbell state it 'enquires into the participant's habitual observable **social behavior**'. • Active scale-**social behaviours** that could be performed without the ability to mentalise (e.g. shares toys when asked). • Interactive scale-measures **social behaviours** contingent upon mental state insight (e.g. plays hide and seek).
Social Competence (Rosner et al., 2004)	• The child behavior checklist (Achenbach, 1991)	• The CBCL requires parents to list activities pursued by their child and to rate the child's **skill** and participation in these activities and how well the **child gets along with others**. • 2 domains: activity and social Activity domain-rates the number of sports the individual is involved with, frequency of participation, skill in sports and non-sports hobbies, number and quality of jobs/chores the individual does. Social domain-number and degree of participation in clubs and

		organisations, number of close friends, degree of contact with friends, **how well the individual gets along** with family and peers, how well the individual plays or works alone.
Social Cognition-four social cognition processes, all part of social information processing; encoding, cue interpretation, strategy generation and evaluation of consequences. (Leffert & Siperstein, 1996)	• No name given to the assessment methods used, although the description sounds as though it is part of the social information processing skills measure (Tur-Kaspa & Bryan, 1994). • Social behaviour scale, consisting of items from other scales: o 14 items from the sociability/leadership scale of the revised class play scale (Masten, Morison & Pellegrini, 1985). o 13 items from the Aggressive behavior checklist (Achenbach & Edelbrock, 1986). o 13 items from the Social withdrawal and anxious scales-from the child behavior checklist (Achenbach, 1991)	• 24 social vignettes, representing two types of social conflict situations (peer entry and peer provocation). After watching the vignettes the children were asked questions on four areas of social information processing: **encoding** of social cues, their **interpretation** of what happened in the story, what **responses they would generate** if they were in the situation and were then asked to rate three different **consequences** to the end of the scenario. • The three scales used to generate the social behaviour scale all measure different aspects of **social behaviour.**
Social Cognition-Social information processing (Gomez & Hazeldine, 1996)	• Social Information Processing task (Suess, Grossman & Sroufe, 1992).	• Six social vignettes were shown to the children which all showed a social dilemma involving provocation by a peer to another child, or their toys. Children were then asked questions around two aspects of social information processing: **encoding and interpretation of social cues** and **responses to social cues**.
Social Cognition-social perception and generation of social strategies (Leffert, Siperstein & Millikan, 2000)	• No name given to the assessment methods used, although the description sounds as though it is part of the social information processing skills measure (Tur-Kaspa & Bryan, 1994).	• Short social vignettes are presented which show social dilemmas. The child is then asked questions to elicit whether they had encoded the social conflict and if they could interpret the intentions of the child in the vignette (**social perception**) and what their response would be in such a situation (**social strategy generation**). Taken as part of the social information processing skills measures, this study could also be interpreted as measuring steps

		of the **social information processing** model: Encoding social cues; representing/interpreting social cues; enactment process.
Social Cognition-Social information processing (Tur-Kaspa , 2004)	• Social information processing skills measure-adapted for use with learning disability populations (Tur-Kaspa & Bryan, 1994).	• Short social vignettes are presented which are about social dilemmas and questions are then asked on 6 areas of **social information processing:** Encoding social cues, representing/interpreting social cues, clarifying goals, searching for possible social responses, making a response decision and enactment process (after Dodge, 1986).
Social Cognition-Social information processing (Bauminger et al., 2005)	• Modified version of the Social Information Processing skills measure (Tur-Kaspa & Bryan, 1994) • The emotion comprehension task (Cermele, Ackerman & Izard, 1995). • The affective matching measure (Feshbach, 1993). • The Kusche affective interview (Kusche, Greenberg & Beilke, 1988)	• Short social vignettes (4) are presented and then questions are asked on 6 areas of **social information processing:** Encoding social cues, representing/interpreting social cues, clarifying goals, searching for possible social responses, making a response decision and enactment process (after Dodge, 1986) • Ability to recognise the **mental state of others** from stories, with social context. • Ability to recognise the **mental state of others** from pictures, with stories. • Key dimensions of **emotional knowledge**: emotional vocabulary, experience of emotions, clues to recognising emotions in oneself and in others, mixed emotions, hiding emotions.
Social Cognition-Theory of mind (Cornish et al., 2005a)	• Location change false belief task • Four appearance-reality tasks	• Measure whether children have **mental state understanding of others** • Measure a child's ability to **understand their own mental state**

Social Cognition-not otherwise specified (Cornish et al., 2005b)	• Facial expression recognition test • Revised eye test • Autism quotient	• Ability to judge **simple mental states** (e.g. happiness, sadness, disgust) from full pictures of facial affect • Ability to judge **complex mental states** (e.g. panicked, jealous, arrogant) from pictures of eyes • 50 statements regarding **social functioning**
Social Skills-social interaction (Kemp & Carter, 2002)	• No official measure, observations based on Carter, Kemp & Iacono (1995). • Social competence ratings-no standardised measure	• Observing the child interacting with other peers with disabilities, without disabilities and with teachers. Specific behaviours were recorded, and also whether the interaction was negative. • Overall social competence-parents were asked to compare their child to a same age typically developing child and rate their social competence compared to this child. Teachers were asked to compare the child to an average peer in their class/grade. Parents and teachers were also asked to rate the child on components of social competence (a) interacting with peers; (b) interacting with adults; (c) self-help skills. Rated on a 4 point scale.
Social Skills (Barton & North, 2004)	• Social Skills Rating System (SSRS; Gresham & Elliott, 1990)	• **Social behaviours-**2 subscales: social skills scale (cooperation, assertion, self-control and responsibility) and problem behaviour scale (externalising problems, internalising problems and hyperactivity).
Social Skills (Fussell, Macias & Saylor, 2005)	• Social Skills Rating System (SSRS; Gresham & Elliott, 1990)	• **Social behaviours-**2 subscales: social skills scale (cooperation, assertion, self-control and responsibility) and problem behaviour scale (externalising problems, internalising problems and hyperactivity).
Social Skills (de Bildt, Luteijn, Kraijer, Sytema & Minderaa, 2005)	• Children's Social Behavior Questionnaire (Luteijn et al., 1998; 2000) • Vineland Adaptive Behaviour Scale (Sparrow, Bella & Cicchetti, 1984) • Autism Behaviour Checklist (Krug et al., 1980)	• Measures **behaviours** on 5 subscales: acting out, social contact problems, social insight problems, anxious/rigid, stereotypical. • Measures adaptive behaviour over four domains: Communication (receptive, expressive and written), socialisation (interpersonal relationships, play and leisure time, and coping skills), daily living skills (personal, domestic and community),

		and motor skills (gross and fine). • Rates 57 **autistic behaviours** on 5 dimensions: sensory, relating, body and object use, language, social & self help.
Social Skills (Tse, Hamiwka, Sherman & Wirrell, 2007)	• Social Skills Rating System (SSRS; Gresham & Elliott, 1990) • Child Behaviour Checklist (Achenbach, 1991)	• **Social behaviours-**2 subscales: social skills scale (cooperation, assertion, self-control and responsibility) and problem behaviour scale (externalising problems, internalising problems and hyperactivity). • The CBCL requires parents to list activities pursued by their child and to rate the child's **skill** and participation in these activities and how well the **child gets along with others**. (See Rosner et al., 2004 above)
Social Skills (Agaliotis & Kalyva, 2008)	• Non standardised observations	• Observations of school break during which time children were encouraged to join in free play. Observers were given operational definitions of the **behaviours** to be recorded. Rated nonverbal **interactions**, including nonverbal initiation and response.
Social Behaviour (Costenbader & Keller, 1990)	• Child behaviour checklist (Achenbach & Edelbrock, 1983). • Conners Rating Scales (Goyette, Conners & Ulrich, 1978)	• 118 **behaviour problems** are rated by parents on a likert scale and fall into two domains: internalizing (overcontrolled) and externalizing (undercontrolled). The scale also has measures of **social competence** in three areas: social, activities and school. • 48 item **behaviour problem** scale
Social Behaviour (Luteijn, Jackson, Volkman, & Minderaa, 1998)	• The Children's Social Behavior Questionnaire (CSBQ, developed by the authors for this study)	• Measures **behaviours** thought to be commonly experienced by individuals with a pervasive developmental disorder.
Social Behaviour (Luteijn, Luteijn,	• The Children's Social Behavior Questionnaire	• Measures **behaviours** on 5 subscales: acting out, social contact

Jackson, Volkman & Minderaa, 2000)	(CSBQ, Luteijn et al, 1998) • Child Behavior Checklist (Achenbach, 1991) • The Autism Behavior Checklist (Krug et al., 1980)	problems, social insight problems, anxious/rigid, stereotypical. • The CBCL requires parents to list activities pursued by their child and to rate the child's **skill** and participation in these activities and how well the **child gets along with others**. (See Rosner et al., 2004 above) • Rates 57 **autistic behaviours** on 5 dimensions: sensory, relating, body and object use, language, social & self help.
Social Behaviour (Lund & Merrell, 2001)	• Home and Community Social Behaviour Scales (Merrell & Caldarella, 1999; Robbins & Merrell, 1998)	• 2 subscales: **social competence** (e.g. 'completes chores or other assigned tasks without being reminded'; 'remains calm when problems arise') and **anti-social behaviour** (e.g. 'ignores parents or supervisors'; 'is physically aggressive').
Social Behaviour (Pierce-Jordan & Lifter, 2005)	• Social Behavior Scale (SocBS, developed by the authors for the study) • Developmental Play Assessment-Behavior Scale (DPA-BS, developed by the authors for the study from the PDA, Lifter, 2000)	• This is a behaviour rating instrument used to rate behaviour on four scales: solitary, onlooking, uncoordinated social, coordinated social. It is used when watching recordings of children's social interactions as a way to rate their **social behaviour.** • This scale is used to rate children's **play behaviour** into four scales: unoccupied, object-focus, mastered play, and emerging play.
Social Behaviour (Hartman, Luteijn, Serra & Minderaa, 2006)	• Children's Social Behavior Questionnaire (refined by the authors for the current study) • Child Behaviour Checklist (Achenbach, 1991)	• Measures **behaviours** on 5 subscales: acting out, social contact problems, social insight problems, anxious/rigid, stereotypical. • The CBCL requires parents to list activities pursued by their child and to rate the child's **skill** and participation in these activities and how well the **child gets along with others**. (See Rosner et al., 2004 above)
Social Behaviour (Zion & Jenvey, 2006)	• School Social Behaviour Scales-second edition (SSBS-2, Merrell, 2000) • Home and Community Social Behaviour Scales	• Rating scale used by teachers, that measures behaviour on two scales: s**ocial competence** (positive) and **anti-social behaviour** (negative) • Parental rating scale, measuring behaviour on two subscales:

	(HCSBS, Merrell & Caldarella, 2002) • Emotionality, activity, sociability (EAS) temperament survey for children (Parental and Teacher ratings, Buss & Plomin, 1984)	**social competence** and **anti-social behaviour.** • Measures children **temperament** through four 'dimensions' of temperament: emotionality, activity, sociability and shyness.
Social Behaviour (Ingram, Dickerson-Mayes, Troxell, & Calhoun, 2007)	• The Playground Observation Checklist (Ingram et al., 2007)	• This is an observation **behaviour** coding schedule with 10 operationally defined behaviours. Behaviours are coded as present or absent.

Social Skills

The topic of social skills has been the focus of a large body of research, particularly with respect to the social skills of children and adults with autistic spectrum disorders. As such it does have the benefit of a number of definitions being provided by researchers, unfortunately the quantity does not aid agreement of a single definition. Matson and Wilkins (2007) summarised this position when they commented that the definitions used for social skills are almost as varied as the studies conducted (p. 29). Bielecki and Swender (2004) stated "a universally accepted definition of social skills does not exist but major themes are reflected in the literature" (p. 694)

One of the ways that social skills have been conceptualised is as a combination of a number of behaviours. Bellack (1983) saw social skills as observable and measurable interpersonal behaviours that promote independence, social acceptability and quality of life. Hughes and Sullivan (1988) discussed a combination of motor, cognitive, and affective behaviours in amounts that would be viewed as neither excessive nor deficient to settings, individuals and/or situations. Whilst such a definition is broad, other researchers have applied even looser descriptions, for example discussing social skills in the context of interactions, communication and play (Wing, Leekam, Libby, Gould, & Larcombe, 2002). Matson and Wilkins (2009) acknowledge that the majority of researchers refer to the interpersonal context and some aspects of reciprocal verbal and non-verbal interactions with at least one other person as a way to understand social skills.

However, there does seem to be a theme of the development of observable behaviours for researchers to use when considering social skills, indeed this is how the majority of social skills assessments are designed. Lushey and Heflin (2000) suggested that providing the definitions of the skills that the research is interested in would be advantageous e.g. 'asking for objects' or 'getting the attention of another'. This is an approach that has been taken in the applied behaviour analytic literature where interventions are based around, often just one, observable, operationalised and well defined behaviour (e.g. spontaneous social exchange, Krantz &

McClannahan, 1998; social initiation, Shabani, Katz, Wilder, Beauchamp, Taylor, & Fischer, 2002; eye contact, Hall, Maynes & Reiss, 2009) Of course, to provide a description for every social skill would be a vast task, but on an individual study level this could be achievable and would allow other researchers to use the operationalised definitions to replicate the research.

A way to reconcile the broader definitions, the need for operationalised behaviours and a definition for social skills per se was offered by Gesten, Weissberg, Amish, and Smith (1987). These authors discussed social skills in relation to social competence and proposed that "social skills are highly specific patterns of learned observable behaviour, both verbal and non-verbal, through which people meet their needs, avoid unpleasant circumstances and influence others" (p.27). Gesten et al., (1987) also suggested that within social skills there are macro-skills (e.g. engaging in conversation, relationship building) and micro-skills (e.g. establishing and maintaining appropriate eye contact). Within this broad definition that provides for both basic and subtle (macro and micro) social skills, there would be the possibility of researchers stating the specific observable behaviour that they were investigating.

Social Skills and Children with Intellectual Disabilities

The variety of definitions in the wider social and cognitive psychology literature is also evident in the intellectual disabilities research. Six papers were identified that matched the search criteria (Kemp & Carter, 2002; Barton & North, 2004; Fussell, Macias & Saylor, 2005; de Bildt, Luteijn, Kraijer, Sytema & Minderaa, 2005; Tse, Hamiwka, Sherman & Wirrell, 2007; Agaliotis & Kalyva, 2008) and four of these (Kemp & Carter, 2002; Barton & North, 2004; deBildt et al., 2005; Agaliotis & Kalyva, 2008) provide definitions for social skills, the particular aspect they are researching or provide comments towards a definition.

Barton and North (2004) give a definition consistent with the concept described by Gesten et al., (1987) "social skills are socially acceptable learnt behaviours considered to be important by others, such as starting a conversation with others or giving a complement" (p. 553). In this

definition the authors have not only specified that social skills comprise a set of behaviours, but have also given some examples for other researchers. Agaliotis & Kalyva (2008) give a similar conceptualisation "social skills represent a specific behaviour that people exhibit in specific situations in order to perform competently on social tasks" (p. 2).

Kemp & Carter (2002) do not provide a definition of social skills, but do provide one for the specific social skill they are investigating, namely social interaction. These authors measure social interaction and as such state that "communicative exchange (verbal or non-verbal); attempts to direct communication to another individual; joint cooperative activity involving two or more individuals; physical actions deliberately directed towards another individual" (p. 397; after Carter et al., 1995, p.22) are all acts of social interaction to be recorded.

Whilst deBildt et al., (2005) do not define social skills per se, they do suggest that there is a difference between basic and subtle social skills "understanding the social context of a situation, understanding jokes, taking the other person's perspective, understanding that a friendly acting person actually is doing you harm, are all examples of subtle social skills needed to handle more complex social situations" (p.318). This discrimination between basic and subtle skills is in accordance with the ideas postulated by Gesten et al., (1987) and are also in line with the recent move in the United States (American Association on Mental Retardation, 2002) towards a definition of intellectual disabilities as requiring the deficit of basic and subtle social skills, such that individuals show certain characteristics e.g.naivety and gullibility.

It can therefore be seen that the concept of social skills is perhaps better defined in the intellectual disabilities literature than social cognition or social competence. This may have been encouraged by the design and application of social skills interventions for children seen as having deficits in the area. It appears that the research already has a number of similar and well accepted definitions for the concept.

The way in which social skills have been measured appears to be consistent with the given definitions. A common measures used across studies was the Social Skills Rating System (SSRS; Gresham & Elliott, 1990) which was employed in three of the six studies. This evaluates social behaviours and has a social skills subscale, comprising cooperation, assertion, self-control and responsibility. The measures employed typically measure social behaviours which are then categorised as social skills. However, the measures seem consistent with the definitions employed in the intellectual disabilities literature.

Social Behaviour

Similar to social cognition, at first glance the term social behaviour would lead us to believe that such a 'common place' term would be easy to define, particularly given its prominence in the other concepts reviewed. However, a search of the literature reveals the opposite is true and even a dictionary definition of the concept is difficult to find. One online dictionary reports the definition "behavior directed towards, or taking place between, members of the same species" (Social behaviour definition, n.d.), which does not acknowledge the social aspects of behaviour. Even books such as 'Handbook of cross cultural psychology: Social behavior and applications' (Berry, Segall & Kagitçibasi, 1997) do not include chapters on social behaviour or define the concept which the book purports to document. It is hard to believe that there is no accepted definition for social behaviour given the emphasis on interventions that have been developed to improve the social behaviours of children with various developmental difficulties.

Perhaps the difficulty lies in the fact that there are so many areas of social behaviour, for example, aggression, assertiveness, altruism, friendship, sharing, cooperation etc., that it is impossible to define the over-arching concept of 'social behaviour'. However, it is important to have at least a broad, rudimentary definition of social behaviour agreed by researchers, as it appears within and referenced by the other concepts which fall under the heading of sociability. If this concept has no agreed definition, it leaves the sociability literature in a difficult position.

Social Behavior and Children with Intellectual Disabilities

The lack of a definition for social behaviour found in the wider literature is replicated in that of children with intellectual disabilities. Eight papers were identified that matched the inclusion criteria and none provided a definition of the concept. However, one of the aims of this paper is to try to assemble definitions and to help in providing working definitions for the main concepts. Therefore, a way of trying to build a definition of social behaviour may come via examining the issues discussed within the papers and the measures used.

Two of the papers highlight difficulties with the construct not being widely defined, with particular reference to a clinical population for which social behaviour problems are central. Hartman, Luteijn, Serra & Minderaa (2006) point out the problem of not having well defined social-behavioural descriptions for children suspected of having pervasive developmental disorder not otherwise specified (PDDNOS). However, until a definition for social behaviour can be agreed upon, descriptions of the specific behaviours will also be difficult to operationalise, therefore impacting on the ability for researchers to agree definitions for social behaviours relevant to PDDNOS. Similarly, Luteijn, Luteijn, Jackson, Volkman & Minderaa (2000) call for an instrument which is reliable and valid, that will describe the social-behavioural problems of children with PDDNOS. These authors developed a questionnaire to address the problem. Pierce-Jordan and Lifter (2005) also discuss a need for researchers to differentiate between play behaviour and social behaviour, and suggest that a number of measures confuse the two aspects. The problems highlighted in the research suggest a need to define not only the concept of social behaviour, but also provide definitions of specific social behaviours. Perhaps this should be done on a study by study basis which would be advantageous in the literature.

The papers use a number of measures purported to measure children's social behaviour. However, some of the measures also include, or focus on, other areas of sociability. Three of the papers use the Child Behavior Checklist (Achenbach & Edelbrock, 1983; Achenbach, 1991)

which is a commonly used measure of behaviour problems. However, as can be seen in the review on social competence, this scale is also used to assess social interaction and a child's ability to interact with other people. This scale can therefore be seen as combining a number of concepts, including social behaviour, social skill and social competence. Other measures rate social competence (School Social Behavior Scales-Merrell, 2000; Home and Community Social Behavior Scales-Merrell & Caldarella, 2002), problem behaviours (Conners Rating Scales-Goyette, Conners & Ulrich, 1978), anti-social behaviour (Home and Community Social Behavior Scales -Merrell & Caldarella, 1999; Robbins & Merrell, 1998; School Social Behavior Scales-Merrell, 2000), temperament (Emotionality, Activity, Sociability (EAS) Temperament Survey for Children (Parental and Teacher Ratings) Buss & Plomin, 1984), autistic behaviours (The Autism Behavior Checklist-Krug, Arick & Almond, 1980), play and play ground behaviour (Developmental Play Assessment-Behavior Scale, Pierce-Jordan & Lifter, 2005; the Playground Observation Checklist, Ingram, Dickerson-Mayes, Troxell, & Calhoun, 2007) and behaviours specific to PDDNOS (the Children's Social Behavior Questionnaire, Luteijn et al., 1998; Luteijn et al., 2000).

Taking an overview of how social behaviour is measured it can be seen to be an all encompassing social term, which includes measures of concepts (social competence) in which social behaviour is referred to; whilst at the same time some measures include detailed descriptions of specific behaviours thought to be 'social' (e.g. the playground observation checklist). This therefore leaves the concept of social behaviour poorly defined.

DISCUSSION

The primary aim of this review was to define and differentiate the constructs used to assess sociability in children with intellectual disabilities (as found in the current literature); and to compare these to definitions of the constructs within social and cognitive psychology literature. Reviewing the social and cognitive literature revealed a number of difficulties with definitions of

the concepts related to sociability, and the difficulties continued with research investigating these social concepts in children with intellectual disabilities.

The literature search revealed four main constructs which are included under the heading of sociability: social cognition, social competence, social skills and social behaviour. In the wider literature the concept of social cognition has received a vast amount of research interest and yet there is no agreed definition. The review suggested that this is perhaps because social cognition is not a unitary concept, but rather an approach or philosophy. However, definitions were found, the main tenets of which coincided with those found in the intellectual disability research and the measures employed.

Social competence was also found to be a wide area with definitions ranging from prescriptive, in terms of stating which dimensions should be measured, through to being extremely flexible and open to interpretation e.g. being well liked (Hubbard and Coie, 1994). No definitions were found in the intellectual disabilities literature although the measures employed to investigate the area all contain specific social behaviours, grouped into areas of social skills deemed necessary for social competence and these dimensions did compare favourably to the definitions suggested in the wider literature.

The conceptualisation of social skills was remarkably uniform across the wider and intellectual disabilities literature, again however, no agreed definitions appear to exist. There is a general theme of conceptualising social skills as a set of observable behaviours which are applied in the appropriate way in certain contexts. The measures employed also reflect this as the majority are scales assessing specific social behaviours.

Unfortunately the area of social behaviour, to which the other three concepts all refer, is extremely poorly defined in both the wider and intellectual disabilities literature. It is suggested that perhaps no definitions exist due to the vast number of behaviours which could be

conceptualised as 'social'. A review of the measures employed to assess social behaviours also highlights the inextricable link between social behaviour and the other three areas of sociability.

Potential working definitions

For social cognition one definition suggested in the general literature was "the perception of others, the perception of self, and interpersonal knowledge" (Beer & Ochsner, 2006). A more detailed definition has been proposed by Bauminger et al., (2005)

> Social cognition includes the child's ability to spontaneously read and correctly interpret verbal and nonverbal social and emotional cues; the ability to recognize central and peripheral social and emotional information; the knowledge of different social behaviours and their consequences in diverse social tasks (e.g. how to initiate a conversation, how to negotiate needs, how to make group entry); and the ability to make an adequate attribution about the other person's mental state (i.e. "theory of mind" abilities or role taking abilities). (p. 45).

Other researchers defined the specific aspect of social cognition they were investigating, and combined with a general definition of social cognition, such as that proposed by Bauminger et al., (2005) this could be seen as useful as it would provide a way for researchers to have a shared, broad concept for social cognition and then specific definitions for the area which is under investigation.

The area of social competence was markedly lacking definitions, although a general definition suggested in the wider literature was "possessing and using the ability to integrate thinking, feeling and behavior to achieve social tasks and outcomes valued in the host context and culture" (Topping et al., 2000, p. 31). In the intellectual disabilities literature no definitions were suggested, although many issues were raised by authors. The suggestion made from reviewing the literature would be to view it as a broad term, encompassing other social concepts (e.g. social

skills and behaviour) with an overarching theme of being able to interact with other people, which allows successful social outcomes that are pertinent to the context the individual is in. It would then be advantageous if researchers defined the specific skills, behaviours etc. they believe necessary for social competence and that they will measure.

As the area of social skills is so vast, it is hardly surprising that a number of definitions exist for the concept. In the wider literature there is an emphasis on a set of behaviours, "social skills are highly specific patterns of learned observable behaviour, both verbal and non-verbal, through which people meet their needs, avoid unpleasant circumstances and influence others" (Gesten et al., p.27). This is reflected in the definitions suggested in the intellectual disabilities research and two possible working definitions which are very similar were suggested by Barton and North (2004) "social skills are socially acceptable learnt behaviours considered to be important by others, such as starting a conversation with others or giving a complement" (p. 553) and Agaliotis & Kalyva (2008) "social skills represent a specific behaviour that people exhibit in specific situations in order to perform competently on social tasks" (p. 2). As with social competence, it would be still remain for individual researcher to state which social behaviours they were interested in investigating in their conceptualisation of social skills.

As discussed above, the area of social behaviour has proven to be somewhat problematic in terms of definitions. It appears as though meaning is implicit such that definitions are seen as unnecessary. The measures employed in the area also do not aid the definition of the concept as many of the measures assess and refer to other concepts within the sociability domain, such as social competence and areas of social cognition. At this stage it is not possible to suggest a working definition for social behaviour, even such a broad definition such as that provided in a dictionary ("behavior directed towards, or taking place between, members of the same species", Social behaviour definition, n.d.). The inherent problem with such a vague notion would be the

ability to construe any behaviour as social. Therefore it is suggested that until research efforts are focused upon defining exactly what is meant by social behaviour, no definition can be provided.

Differentiating the constructs

One of the aims of this review was to provide a way to differentiate the four main social concepts from each other; however, a review of the definitions reveals that this will not be a simple task. If the suggested working definitions are utilised then it could be seen that social competence is an overarching concept that includes facets of social cognition, social skills and social behaviour. Such that someone possessing good social cognition and social skills (including social behaviour) would be classed as 'socially competent'. Social cognition can then be seen as encapsulating social skills and social behaviours; with social skills requiring a specific set of (undefined) social behaviours. However, numerous questions remain which have been outside of the remit of this review, such as: can someone possess social skills without social cognition? Can someone have adequate social behaviours but still be seen as not socially competent? Can social skills be 'taught' to someone if they lack social cognition?

It would appear that the concepts are to some extent inextricably linked and both definitions and differentiations will take concerted efforts to achieve.

Clinical Implications

The need for clinical interventions for children with intellectual disabilities who have sociability problems has long been recognised and numerous interventions have been designed and implemented. The majority of interventions are targeted at social skills deficits for children with autism or autistic spectrum disorders. However, as discussed in the previous sections, the lack of coherent definitions for social skills and the behaviours pertinent to social skills makes it a difficult area to conceptualise. Therefore, designing and implementing interventions for a social concept which is not universally agreed can have many implications for researchers and individuals in the social skills programmes.

In a recent review of social skills training programmes for children with high functioning autism or Aspergers disorder, it was found that 70% of programmes reported success (Rao, Beidel, & Murray, 2008). However, the authors point out that within these successful programmes, success was only for a subset of children or on a subset of skills. The first limitation Rao et al., (2008) discuss is the lack of a universally accepted definition of social skills and social behaviours thought to be deficient in the children, and therefore requiring intervention.

Thus, the implications of not having well defined and differentiated concepts are not only a problem for the research community but also for clients of clinical services. The impact of this problem is therefore extremely serious and far reaching. Without clear definitions of behaviours and skills that need targeting, how can outcome and success be measured?

Future Directions

It was anticipated that one of the outcomes of this review would be to add to the current definitions employed in the literature in a bid to improve cohesion throughout the research arena. However, the lack of definitions (e.g. for social behaviour) or the wide variety of conceptualisations (e.g. social cognition and social competence) make such a task extremely difficult at the present time. Whilst some potential working definitions have been suggested, future efforts should be focused on trying to bring together the diverse ideas around some of these concepts so that working definitions can be debated and implemented in future research.

At the very least researchers interested in aspects of sociability should be aware of the lack of definitions currently available and provide their own idiosyncratic definitions as a minimum standard. Further, clinicians implementing social interventions for children with social deficits need to be aware of the lack of definitions of the concepts they are aiming to treat. The implications for assessment, formulation and intervention are far reaching. Without clear definitions: how can assessments be judged as reliably measuring the behaviours and skills a child is thought to be deficient in? How can formulation truly capture the nature of a child's

difficulties if no definition exists for the difficulty purported to be experienced? How can interventions target a deficiency and be measured for effectiveness if no universally accepted definitions exist?

Whilst this review has perhaps raised more questions than answers, without consideration of such fundamental principles such as clear definitions the conclusions drawn from research need to be carefully considered.

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2. Empirical Paper

Delineation of a Behavioural Phenotype for Monosomy 1p36 Deletion Syndrome: Autism, Affect, Hyperactivity and Sociability

ABSTRACT

Background: Research on Monosomy 1p36 deletion syndrome indicates there may be behavioural characteristics associated with the condition. However, there is no specific research on the social and behavioural phenotype of the disorder. The primary aim of this study is to delineate the behavioural phenotype for the condition, with a particular emphasis on the social phenotype by comparing individuals with Monosomy 1p36 to matched individuals with three other genetic syndromes (Angelman, Cri du Chat, and Cornelia de Lange) on measures of social behaviour and to observe social behaviour in experimental social presses.

Method: 90 participants were included in the comparison study, aged between eighteen months and forty five years. Twelve individuals aged between three years three months and thirteen years eleven months who had a confirmed diagnosis of Monosomy 1p36 deletion participated in the observation study. A number of behavioural measures were employed and individuals were observed interacting with a familiar and unfamiliar adult where adult engagement/attention was manipulated across five conditions. Video recordings of the observations were coded for social behaviours and skills.

Results: Results from the comparative study indicate impaired social communication, lowered mood and higher sociability with familiar adults are all notable characteristics for 1p36. In the social presses, individuals were more social under conditions of high attention/engagement with both familiar and unfamiliar people.

Conclusions: The study is the first to investigate social behaviour in 1p36 syndrome and as such the conclusions drawn are tentative. There is evidence that some

characteristics may form part of a behavioural and social phenotype for the condition.

INTRODUCTION

Monosomy 1p36 Deletion Syndrome

Monosomy 1p36 deletion syndrome is the most common terminal deletion with an estimated incidence between 1 in 5,000 (Heilstedt, Ballif, Howard, Kashorf, & Shaffer, 2003) to 1 in 10,000 live births (Shapira, McCaskill, Northrup, et al., 1997). Approximately 150 case descriptions have been published (Battaglia, Hoyme, Dallapicolla, et al., 2008) since the syndrome was first identified (Hain, Leversha, Campbell, Daniel, Barr, & Rogers, 1980). The syndrome results in a number of clinically identifiable characteristics, including craniofacial, skeletal, visceral, neurological and developmental features. 1p36 syndrome can be recognised and diagnosed by distinct facial dysmorphology which characterises the disorder, which include, but are not restricted to: large, late closing anterior fontanelle, microcephaly (small head), brachycephaly (flatness across the back of the head), frontal bossing (prominent forehead), deep set eyes, flat nasal bridge/nose, pointed chin, small palpebral fissures, and low set abnormally formed ears (see Table 2.1 for a review of typical features in the syndrome).

A variety of medical problems are associated with the condition, the majority of which can be treated. The most common disorders are heart problems, with structural heart defects reported in up to 75% of individuals (Gajecka, Mackay & Shaffer, 2007). Epilepsy is also found in the majority (Slavotinek, Shaffer & Shapira, 1999) and can be well controlled by medication (Battaglia et al., 2008). Feeding difficulties are often evident in infancy, with some children requiring a temporary nasogastric tube (Heilstedt, et al, 2003). In a review of 134 published cases of 1p36 syndrome, Gajecka, et al., (2007) report high prevalence of gastrointestinal problems. The most common of these were constipation (65%) and reflux (56%).

Both visual and hearing impairments are also consistently observed in approximately 50% of participants (Battaglia et al., 2008; Gajecka et al., 2007). Visual inattentiveness is noted (Battaglia

et al, 2008; Gajecka et al., 2007, Heilstedt et al., 2003) however, given the large proportion of patients who have definable ophthalmic conditions (e.g. strabismus, myopia, refractive errors) it is difficult to establish whether this is a discrete disorder or a consequence of other visual impairments.

Intellectual impairment is present in all individuals with Monosomy 1p36-ranging from mild to profound (Shapira et al., 1997; Gajeka et al., 2007), with approximately 85% of individuals in the severe to profound range (Battaglia et al, 2008; Shapira et al., 1997) and development typically delayed for all milestones (Battaglia et al., 2008). Speech delay is also common with expressive language severely impaired or absent (Battaglia et al., 2008).

The Social and Behavioural Phenotype of 1p36 Syndrome

Whilst the physical phenotype of the syndrome has been characterised, little emphasis has been placed on the behavioural and social phenotype. However, research on other genetic syndromes has begun to investigate distinctive social traits. Most notably, research on Williams and Fragile X syndromes has focused on the prominent and markedly different social abnormalities observed in the conditions. The early study of the social phenotypic traits of these two disorders has created a precedent for investigating social traits in other genetic syndromes (Feinstein & Singh, 2007).

There are some indications of particular social traits associated with 1p36 syndrome (e.g. Battaglia, 2005; Battaglia et al., 2008); however, the lack of psychometrically robust psychological assessment of these characteristics makes it difficult to draw conclusions. Behaviourally the syndrome appears to have some phenotypic traits, including self-injury and temper tantrums; again, however, behaviour is not regularly reported in the literature and a paucity of standardised adaptive behaviour measures makes it difficult to compare the syndrome with normative data or with other syndrome groups.

Table 2.1: Common features of 1p36 syndrome as reported in 11 papers. If a feature is not reported, it does not indicate the feature does not exist within the population described, it may not have been assessed for.

	Paper											
Feature	1[1]	2[2]	3[3]	4[4]	5[5]	6[6]	7[7]	8[8]	9[9]	10[10]	11[11]	12[12]
Craniofacial												
Microcephaly	+	+	+	+		+	+	+	+			+
Brachycephaly	+			+	+	+	+	+				+
Large, late closing anterior fontanelle	+	+	+	+	+	+	+				+	+
Abnormal ears (small, low set, posteriorly rotated)	+	+	+	+	+	+	+		+	+		+
Deep set eyes	+			+	+	+	+			+	+	+
Small palpebral fissures	+		+	+	+	+	+					
Epicanthal folds		+	+		+				+		+	
Flat nasal bridge	+	+	+	+	+	+	+			+	+	+
Pointed chin	+	+	+	+	+	+	+		+		+	
Midface hypoplasia	+	+	+			+						+
Straight eyebrows	+	+	+						+			+
Forehead bossing			+		+	+			+	+		+
Medical												
Gastrointestinal anomalies (ulcer, constipation, reflux, hernia)	+	+			+							
Congenital heart defects	+	+	+	+	+	+	+		+			+
Brachydactyly/ camptodactyly / clinodactyly	+	+	+	+	+	+	+		+			+
Abnormalities of external genitalia		+		+	+	+						
Skeletal abnormalities		+			+							
Small hands and feet		+	+	+	+							+
Growth delay				+	+							
Obesity				+	+	+						
Renal abnormalities		+			+							
Hyperthyroidism			+			+	+					
Vision/hearing												
Hypermetropia	+					+	+					
Myopia	+						+					
Strabismus	+	+		+	+	+	+					+
Visual inattentiveness	+	+	+	+		+	+					+
Refractive errors		+			+	+						
Conductive hearing loss	+	+		+	+	+	+					+
Sensorineural hearing loss	+	+	+	+	+	+	+					+
Neurological/developmental												
Intellectual impairment	+	+	+	+	+	+	+	+	+	+	+	+
Developmental delay	+	+	+	+	+	+	+	+	+	+	+	+
Speech delay	+	+	+						+			
Self-injurious behaviour	+	+	+	+	+	+					+	
Autistic features		+	+		+					+	+	+
Temper tantrums		+			+							
Reduced social interaction		+									+	
Poor eye contact										+	+	
Stereotypies		+									+	
Repetitive behaviour		+									+	
Hypotonia	+	+	+	+	+	+	+	+	+	+	+	+
Epilepsy/seizures	+	+	+	+	+	+	+		+	+		+
Feeding difficulties	+			+			+	+	+	+	+	+
Oropharyngeal dysphasis	+		+			+	+					

[1] Gajecka, Mackay & Shaffer (2007); [2] Battaglia et al., (2008); [3] Battaglia (2005); [4] Shapira et al., (1997); [5] Slavotinek, Shaffer & Shapira (1999); [6] Slavotinek (2003); [7] Heilstedt et al., (2003) [8] Rudnik-Schöneborn, Häusler, Krings, & Schüler (2008); [9] Kang et al., (2007); [10] Blennow, Bui, Wallin & Kogner (1996); [11] Tong, et al., (2005); Knight-Jones et al., 2000.

The problematic behaviours commonly reported in the literature are aggression, temper tantrums, throwing or banging objects, striking people, screaming, self-injury and autistic features. Typically, the focus of research has been on the clinical phenotype and clinical or molecular characterisation of the syndrome and has therefore paid less attention to behaviour. In nine out of eleven published articles (see Table 2.1) aspects of behaviour are mentioned, with only three of these providing some description of the behaviour (Battaglia, 2005; Battaglia et al., 2008; Slavotinek, et al., 1999). No comparisons are made with individuals of similar chronological or developmental ages, level of ability or with other genetic syndromes and this is considered critical to identification of a behavioural phenotype (Dykens and Hodapp, 1999).

Of the behaviours which are recorded, self-injury is the most widely reported and detailed. Prevalence rates of self-injurious behaviour vary across studies from 30% (Battaglia et al, 2008), to 56% (Gajecka et al., 2007; Shapira et al., 1997). These can be compared to prevalence rates suggested in general learning disability populations (10-50% life time prevalence, Borthwick-Duffy, 1994); Prader-Willi syndrome (81%, Symons, Butler, Sanders, Feurer, & Thompson, 1999); Smith-Magenis syndrome (between 92% Arron et al., 2008 & 98%, Dykens & Smith,1998); Fragile X syndrome (58%, Symons, Clark, Hatton, Skinner & Bailey, 2003) and Cornelia de Lange syndrome (55.6% Oliver, Sloneem, Hall, & Arron, in press).

The commonly detailed self-injurious behaviour in 1p36 deletion syndrome consists of hand biting, wrist biting, head striking/banging, and sucking fingers excessively. Less commonly reported behaviours are self-pinching and scratching the peroneal region on the foot (Battaglia, 2005). Whilst self-injury is reported in the literature, it has not been a focus of investigation and therefore no attempts to ascertain why it occurs have been made. However, given the prevalence of medical difficulties associated with the condition, including gastrointestinal anomalies (see also Table 2.1), it is possible that some of the observed self-injury is associated with pain and

discomfort (Luzzani, Macchini, Valade, Milani, & Selicorni, 2003). At present this is a much neglected aspect of the syndrome and is currently being researched (Marr, 2009).

As well as research neglecting behavioural aspects of 1p36 syndrome, social aspects of the condition have also not been described in detail. Of eleven papers reviewed, six discuss social aspects, including behaviour (Blennow, The-Hung, Anders & Per, 1996; Slavotinek, et al., 1999; Battaglia, 2005; Tiong, et al., 2005; Knight-Jones et al., 2005; Battaglia et al., 2008). However, only three of these papers describe the difficulties, whilst six note 'autistic features' (Blenow et al., 1996; Slavotinek et al., 1999; Battaglia, 2005; Knight-Jones et al., 2005; Tong et al., 2005; Battaglia et al., 2008). The difficulties described, which are indicative of both social difficulties and/or autism are: reduced social interaction, repetitive stereotypies (e.g. holding hands in front of face, hand washing or flapping), tendency to beat, smell or roll objects repetitively, manual apraxia (twisting hands in a washing manoeuvre) and poor eye contact (Blennow et al., 1995; Tiong et al., 2005; Battaglia et al., 2008).

As social aspects have not been the focus of research efforts, no consistent assessments or methods of obtaining information on sociability in the syndrome have been applied. Some studies do not report any social traits or difficulties, so a concerted effort to apply a consistent approach to the area is required. This will be best achieved through using standardised methods of assessing sociability that have been adapted for use with individuals with intellectual disability.

When standardised assessment methods are applied to the syndrome it will be possible to understand the social aspects of the condition with reference to other known genetic syndromes. Comparisons between syndromes known for excessive sociability (e.g. Williams's syndrome; Jones et al., 2000; Angelman Syndrome; Oliver, Berg, Moss, Arron & Burbidge, in press) or excessive social deficits (e.g. Cornelia de Lange syndrome; Moss, Kaur, Jephcott, Berg, Cornish & Oliver, 2008; Oliver, Arron, Hall & Sloneem,2008; Richards, Moss, O'Farrell & Oliver, in press) can then be made. These comparisons begin the process of delineating the social phenotype of

the condition. It is therefore important that comparable groups and participants are chosen primarily based on level of ability.

There is a difficulty, however, with only employing standardised assessment measures, as these may not capture the nuances of sociability or social interactions. Therefore, methods that can measure and categorise sociability into the different elements that may constitute it (e.g. social communication skills, social motivation); and which can systematically alter the conditions under which social traits may be expressed, may provide a rich source of information about sociability in 1p36 syndrome. Such a method combined with standardised assessments may provide a good starting point from which to begin to characterise the social phenotype of the syndrome.

Aims of the Current Research

The primary aim of the current research is to begin the process of characterising the behavioural phenotype with a focus on the social phenotype for Monosomy 1p36 deletion syndrome and compare this to other genetic syndromes (Angelman syndrome, Cornelia de Lange syndrome and Cri du Chat syndrome). By employing measures that are appropriate, reliable and valid for individuals with intellectual disabilities; and incorporating a structured social press paradigm to assess sociability, the research will extend existing literature.

STUDY ONE

The first study is a comparison between a group of individuals with 1p36 deletion syndrome and comparable individuals from three other genetic syndrome groups (Angelman, Cornelia de Lange and Cri du Chat) on a number of measures of social behaviour and sociability (including mood, repetitive behaviour, social communication, hyperactivity, impulsivity and familiar and unfamiliar social interaction) using standardised questionnaires. The aim of this study is to compare individuals with 1p36 syndrome to other genetic syndromes where behavioural and social phenotypes have already begun to emerge.

As there is no agreed upon definition of sociability the current research employs a working definition of the relative tendency or disposition to be sociable or associate with other people and acknowledges that such concepts as social cognition, behaviour, skills and competence are all aspects of sociability.

METHOD

Participants

Recruitment

Participants with 1p36 syndrome were recruited via the Unique Rare Chromosome Disorder Support Group and parents were asked to complete a questionnaire pack. This group provides support for families affected by a number of very rare chromosome disorders. 54 families affected by a pure 1p36 deletion were registered with the group at the time of the research. Only individuals who had a diagnosis of a 1p36 deletion with no other genetic or chromosomal disorder from a General Practitioner, Clinical Geneticist, Paediatrician, Neurologist or Psychiatrist were included. Data were excluded if more than 25% of information from any questionnaire was missing. 54 packs were sent out and 26 were returned (48.14% return rate). After applying inclusion and exclusion criteria the overall return rate was 42.6% (23). Unique sent questionnaire packs (covering letter, information sheet, questionnaires, consent form and prepaid return envelope, see appendix 1a) to carers of potential participants. They were asked to complete and return the questionnaires and consent forms if they wished to take part in the study.

Participants with three other genetic syndromes (Angelman (AS), Cornelia de Lange (CdLS) and Cri du Chat (CdCS) had previously taken part in a large scale behavioural phenotype study (Oliver, et al., in review) and had given consent for their information and data to be included in future studies.

Participant Information

Table 2.2 details the participant information. The 23 participants in the 1p36 syndrome group were matched with individuals from an existing database. The matched participants had one of three syndromes: Angelman, Cornelia de Lange and Cri du Chat. Participants were matched on two variables from the Wessex Scale (see below), verbal ability and self-help score (+ or – 2 points). 23 individuals were matched from both the Cornelia de Lange and Cri du Chat syndrome groups. However, only 21 individuals could be matched from the Angelman syndrome group.

Across the four groups 90 participants (65.6% female) were included in the study with an age range of 18 months to 45 years (mean 10.35, SD 7.96). Level of ability (self-help skills), mobility (ability to walk unaided), speech, visual and auditory impairment were described by The Wessex Scale (Kushlick, Blunden & Cox, 1973). Overall 17 (18.9%) were able or partly able, 31 (34.4%) were mobile, 42 (46.7%) were verbal (used more than 30 words or signs), 65 (72.2%) had normal hearing and 56 (62.2%) had normal vision.

Table 2.2: Demographic characteristics for each group. (1p36=1p36 terminal deletion syndrome, AS=Angelman Syndrome, CdCS=Cri du Chat Syndrome, CdLS=Cornelia de Lange Syndrome).

		1p36	AS	CdCS	CdLS	Total
N		23	21	23	23	90
Age[1]	Mean	7.29	11.64	11.28	11.29	10.35
	(SD)	(4.19)	(10.49)	(8.09)	(7.75)	(7.96)
	Range	1.05-16.04	1.98-45.08	1.59-33.06	1.89-29.22	1.05-45.08
Gender	Female	16 (69.9%)	12 (57.1%)	15 (65.2%)	16 (69.6%)	59 (65.6%)
Self help[2]	Partly[3] able/able	4 (17.4%)	2 (9.5%)	5 (21.7%)	6 (26.1%)	17 (18.9%)
Mobility[2]	Fully mobile[4]	10 (43.5%)	4 (19%)	11 (47.8%)	6 (26.1%)	31 (34.4%)
Vision[2]	Normal	8 (34.8%)	18 (85.7%)	20 (87%)	10 (43.5%)	56 (62.2%)
Hearing[2]	Normal	14 (60.9%)	21 (100%)	19 (82.6%)	11 (47.8%)	65 (72.2%)
Speech[2]	Verbal	9 (39.1%)	6 (28.6%)	12 (52.2%)	15 (65.2%)	42 (46.7%)

Measures

Parents and carers completed eleven questionnaires, seven of which were utilised in the current study. The other questionnaires are being reported in other research (Marr, 2009). The seven questionnaires reported are:

Demographic Questionnaire

This detailed age, gender, mobility, verbal ability, diagnostic status (including when the person was diagnosed and by whom).

The Wessex Scale (Kushlick et al., 1973)

This is a carer report measure used to assess social and physical abilities in individuals with an intellectual disability. Subscales cover continence, walking, self-care, vision, hearing, speech, and

[1]In years.
[2] Data derived from the Wessex questionnaire (Kushlick *et al.,* 1973).
[3] Those who score six or above on the total score of the self help subscale (items g-i).
[4] Those who score six or above on the total score of the mobility subscale (items e & f).

literacy. The scale has good inter-rater reliability at subscale level for both children and adults (Kushlick et al., 1973; Palmer & Jenkins, 1982).

Social Communication Questionnaire (SCQ; Rutter, Bailey & Lord, 2003)

Formerly known as the Autism Screening Questionnaire, this is a brief screening instrument to evaluate social and communication skills in children suspected of having an autistic spectrum disorder. Three subscales assess communication, social interaction and repetitive and stereotyped behaviours. Items are scored for the presence or absence of abnormal behaviours: higher scores indicate the presence of more abnormal behaviours. Scores of 15 and above distinguish individuals with an Autistic Spectrum Disorder and 22 and above denotes Autism. The questionnaire shows good concurrent validity with the Autism Diagnostic Interview and the Autism Diagnostic Observation Schedule (Berument, Rutter, Lord, Pickles, & Bailey, 1999; Howlin & Karpf, 2004).

The Activity Questionnaire (TAQ; Burbidge & Oliver, 2008)

This is an informant based questionnaire that assesses hyperactivity and impulsivity in individuals with an intellectual disability. It comprises 18 items grouped into three subscales: overactivity, impulsivity and impulsive speech. Burbidge and Oliver (2008) identified cut-off points for the overactivity (32) and impulsivity (24) subscales to identify individuals scoring abnormally high. Item level inter-rater reliability ranges from .31 to .75 (mean 056) and test-retest reliability ranges from .60 to .90 (mean .75). Inter-rater reliability and test-retest reliability indices for subscales and total score exceed .70 (Oliver et al., in review).

Mood, Interest and Pleasure Questionnaire Short Version (MIPQS; Ross & Oliver, 2003; Ross, Aaron & Oliver, 2008)

An informant based questionnaire comprising twelve items in two subscales: Mood, and Interest and Pleasure and is used to assess these two aspects of depression in individuals with an intellectual disability. The questionnaire has good psychometric properties, with good internal

consistency (Cronbach's alpha coefficients: total=.88, Mood= .79, Interest and Pleasure= .87), test-retest (.97) and inter-rater reliability (.85), (Ross and Oliver, 2003). Cut-off points have been identified (Ross et al., 2008) to indicate individuals scoring abnormally high (23-interest and pleasure; 24-mood) or low (6-interest and pleasure; 15-mood).

The Repetitive Behaviour Questionnaire (RBQ; Moss and Oliver, 2008)

Nineteen item informant based questionnaire for use with individuals with an intellectual disability. Five subscales (stereotyped behaviour, compulsive behaviour, insistence on sameness, restricted preferences and repetitive use of language) assess specific types of repetitive behaviour. The questionnaire has good psychometric properties, with inter-rater reliability for items ranging from .46 to .80, with 73% of items above .80; test-retest reliability coefficients range from .61 to .93 at item level (Moss & Oliver, 2008; Moss, Oliver, Arron, Burbidge & Berg, 2009; Oliver et al., in press) and good convergent validity with the repetitive behaviour subscale of the Autism Screening Questionnaire (Berument et al., 1999).

The Sociability Questionnaire for Intellectual Disabilities (SQID; Collis & Oliver,2007)

A recently developed informant based questionnaire for use with intellectual disability populations. It assesses social interaction with familiar and unfamiliar people. It comprises 25 items in eight subscales (four familiar and four unfamiliar): receive interaction (receiving an interaction/being approached by another), interaction (one-on-one ongoing interaction), approach or initiate interaction (initiating an interaction with another) and performance (a group interaction). No psychometric properties have been established yet, although preliminary analyses (Collis & Oliver, unpublished) have revealed clinical cut off points for subscales for excessive sociability (13 and above) and shyness (below 3).

Data Analysis

The demographic characteristics of the participants were examined using a one-way ANOVA (age) and Chi-squared tests, followed by pairwise post-hoc comparisons. Group differences were

examined using mean scores from each of the four syndrome groups on four questionnaires: The Mood, Interest and Pleasure Questionnaire (MIPQ), The Activity Questionnaire (TAQ) and the Social Communication Questionnaire (SCQ) and the Repetitive Behaviour Questionnaire (RBQ).

Kolmogorov-Smirnov tests of normality were conducted on the subscales and these data were then subject to two sets of analysis. Firstly, one-way ANOVA followed by Scheffé post-hoc analysis (and Kruskal-Wallis followed by Mann-Whitney tests) to investigate group differences on the subscales of the MIPQ, TAQ, SCQ and RBQ. Secondly, Chi-squared comparisons to identify the proportion of each group attaining scores at or above cut-off scores for the SCQ (indicating Autism and Autism Spectrum Disorder), abnormally high and low scores on the Mood and Interest and Pleasure subscales of the MIPQ (indicating positive and negative affect; and low and high Interest and Pleasure), and abnormally high scores on the over activity and impulsivity subscales of the TAQ.

The analyses focus upon differences and similarities between the 1p36 syndrome group and three other comparison groups. Differences between the three comparison groups have been published elsewhere (Arron et al, 2008; Oliver et al., in press; Marr, 2009).

As the SQID is a recently developed questionnaire, no data exist for the comparison groups; therefore analysis of subscales and total scores was completed using pairwise t-tests for the 1p36 group (after completing Kolmogorov-Smirnov tests). Also the proportion of individuals falling above and below clinical cut off points is presented.

RESULTS

Demographic Characteristics

Participant information can be found in Table 2.2 (see page 11). Although the participants were matched on verbal and self-help ability, further analyses were conducted in order to check for differences between groups.

A one way ANOVA revealed no significant difference between the four groups ($F(3) = 1.556$, $p > .05$) on chronological age. Chi-square tests of gender ($\chi^2(3) = .987$, $p => .05$), speech ($\chi^2(3) = 6.748$, $p > .05$), mobility ($\chi^2(3) = 6.364$, $p > .05$) and ability ($\chi^2(3) = 2.136$, $p > .05$; exact test) revealed no significant differences between the participant groups. However, there were significant differences between groups on hearing ($\chi^2(3) = 17.232$, $p < .001$) and vision ($\chi^2(3) = 20.749$, $p < .001$). Post hoc analyses revealed that individuals with Angelman Syndrome and Cri du Chat Syndrome have significantly better vision than individuals with 1p36 Syndrome ($\chi^2(1) = 10.946$, $p < .001$); ($\chi^2(1) = 12.244$, $p < .001$).

Comparisons of Behavioural Differences

MIPQ, TAQ, SCQ and RBQ subscale analyses

The analysis of the four questionnaires will allow a comparison between 1p36 syndrome and the three comparison syndromes on measures of mood, hyperactivity, social communication and repetitive behaviour. Kolmogorov-Smirnov tests revealed one violation of normality in the 1p36 group on the SCQ communication subscale ($p<0.01$) and numerous violations on the RBQ subscales across all groups ($p<.01$). Therefore, mean scores on the SCQ communication subscale were subject to a Kruskal-Wallis analysis indicating a significant effect of group ($\chi^2(3)=13.551$, $p < .001$). A Mann Whitney post hoc test revealed individuals with 1p36 Syndrome scored higher (indicating more impairment) than individuals with Cri du Chat Syndrome ($U = 83.500$, $N_1 = 22$, $N_2 = 20$, $p < .001$).

The RBQ subscales were also subject to Kruskal-Wallis analyses. Only one subscale (restricted preferences) showed a significant effect of group ($\chi^2(3)=9.751$, $p < .05$), with the post hoc revealing the Cri du Chat Syndrome group scored higher (indicating more restricted preferences) than the 1p36 group ($U = 6.500$, $N_1 = 7$, $N_2 = 7$, $p < .01$).

All other subscales were subject to one-way analysis of variance, revealing significant differences on three of the subscales; MIPQ Mood (F (3) =4.242, $p < .01$), SCQ Social Interaction (F (3) =4.094, $p < .01$), and TAQ Impulsivity (F (3) =7.447, $p < .01$). Scheffé post hoc analyses indicated two of the post hoc comparisons were significant for the 1p36 group (see Table 2.3). The Angelman Syndrome group scored higher on the MIPQ Mood subscale than 1p36 group ($p < .01$) and on the TAQ Impulsivity subscale ($p < .001$). All these results can be seen in Table 2.3.

Table 2.3: Means/median# (standard deviations) for subscales of the Mood, Interest and Pleasure Questionnaire, The Activity Questionnaire, the Social Communication Questionnaire and the Repetitive Behaviour Questionnaire with results for analysis of variance and post hoc analyses.

	1p36	AS	CdCS	CdLS	df	ANOVA/ Kruskal-Wallis* F/χ^{2*}	*p*	Post Hoc
N	23	21	23	23				
MIPQ-S Mood subscale	17.77 (3.50)	21.33 (2.51)	19.55 (3.54)	19.90 (3.47)	3	4.242	<.01	AS>1p36 (*p*<.01)
MIPQ Interest & Pleasure subscale	15.72 (5.57)	18.24 (4.09)	17.46 (3.51)	15.26 (4.55)	3	2.145	>.05	
SCQ Communication subscale	8.0# (2.24)	6.0# (1.56)	5.0# (1.98)	6.0# (2.05)	3	13.551*	<.01	1p36> CdCS (*p*<.001)
SCQ Social Interaction subscale	8.56 (3.95)	7.13 (2.32)	5.74 (3.61)	9.26 (3.40)	3	4.094	<.01	CdLS>CdCS (*p*<.05)
SCQ Repetitive Behaviour subscale	3.82 (2.20)	3.45 (1.89)	4.34 (1.30)	3.82 (1.91)	3	0.859	>.05	
TAQ Overactivity subscale	14.57 (10.13)	21.86 (9.41)	18.74 (7.09)	14.40 (7.93)	3	3.730	>.05	
TAQ Impulsivity subscale	9.97 (8.33)	19.07 (5.39)	15.34 (6.33)	11.45 (7.52)	3	7.447	<.001	AS>CdLS (*p*< .01) AS>1p36 (*p*<.001)
TAQ Impulsive Speech subscale	1.71 (1.25)	3.20 (2.17)	3.42 (4.08)	3.00 (2.65)	3	0.522	>.05	
RBQ Stereotyped Behaviour subscale	6# (4.45)	8# (3.53)	9# (4.24)	8# (3.84)	3	3.757*	>.05	
RBQ Compulsive Behaviour subscale[1]	2.25 (3.57)	2.04 (3.17)	2.61 (4.98)	1.09 (3.12)	3	4.846*	>.05	
RBQ Insistence on Sameness subscale	1.52 (2.52)	1.609 (2.46)	1.77 (2.77)	1.68 (2.44)	3	0.035*	>.05	
RBQ Restrictive Preferences subscale	7# (2.85)	6# (1.58)	3# (1.57)	4# (2.43)	3	9.751*	<.05	AS>CdCS (*p*<.01) CdCS>1p36 (*p*<.01)
RBQ Repetitive Use of Language subscale	5# (4.82)	10# (5.37)	3# (2.57)	4# (1.70)	3	3.166*	>.05	

[1] Mean reported the for RBQ subscales compulsive behaviour and insistence on sameness due to low numbers negating the use of the median.

SQID analysis

As can be seen in Table 2.4 individuals with 1p36 syndrome consistently obtained higher scores on the familiar scales of the SQID, indicating more social interaction with familiar people than unfamiliar.

Paired t-tests indicated significant differences between familiar and unfamiliar on each of the four subscales and the total scale scores ($p<.001$).

Table 2.4: Means (standard deviations) for subscales of the sociability questionnaire for intellectual disabilities, for both familiar and unfamiliar social interactions.

Subscale	Familiar	Unfamiliar	T-test	
			t	*p*
Receive interaction	10.91 (2.55)	7.73 (3.29)	4.44	<.001
Interaction	11.54 (1.96)	8 (2.88)	5.77	<.001
Approach or initiate interaction	8.58 (3.37)	6.23 (2.52)	4.82	<.001
Performance	10.58 (2.63)	7.92 (3.40)	4.08	<.001
Total	41.65 (8.61)	29.88 (10.72)	5.16	<.001

Table 2.5 shows the number of individuals falling above and below clinical cut off points on the SQID. Individuals scoring above the cut off point are deemed to show excessive sociability and those falling below, excessive shyness.

Table 2.5: Proportion of individuals with 1p36 falling above and below the clinical cut off points on the SQID.

Subscale	Above clinical cut off	Below clinical cut off
Familiar receive interaction	9 (43.6%)	0 (0%)
Familiar interaction	8 (30.8%)	0 (0%)
Familiar approach or initiate interaction	5 (19.2%)	1 (3.8%)
Familiar performance	6 (23.1%)	0 (0%)
Unfamiliar receive interaction	2 (7.7%)	3 (11.5%)
Unfamiliar interaction	2 (7.7%)	1 (3.8%)
Unfamiliar approach or initiate interaction	1 (3.8%)	3 (11.5)
Unfamiliar performance	2 (7.7%)	1 (2.2%)

Autism and Autistic Spectrum Disorder

The proportion of each group scoring at or above the cut off scores for Autism and Autism Spectrum Disorders (ASD) on the SCQ are shown in Table 2.6. A Chi-square analysis showed no significant differences between group and the proportion of individuals scoring above the cut-off for ASD ($\chi^2(3) = 3.484, p > .05$), but did show a significant difference between group and the proportion of individuals scoring above the cut-off for Autism ($\chi^2(3) = 8.987, p < .05$).

Post hoc analyses were completed using pairwise Fishers exact tests or Chi-square tests. The results of these suggest individuals in the 1p36 are significantly more likely to score above the cut-off for Autism than individuals in the Angelman ($\chi^2(1) = 6.600, p < .01$) or Cri du Chat groups ($\chi^2(1) = 5.301, p < .05$). No other significant differences were found.

Table 2.6: Proportion of each group scoring abnormally low on the MIPQ Mood and Interest and Pleasure subscale; abnormally high on the MIPQ Mood and Interest and Pleasure subscale; abnormally high on the TAQ Overactivity subscale; abnormally high on the TAQ Impulsivity subscale; and at or above the cut off points for autism and autistic spectrum disorder on the SCQ.

	1p36	AS	CdCS	CdLS	Fishers exact/Chi-square* test	Post hoc
MIPQ Mood abnormally low	6 27.3%	2 9.5%	3 13.6%	2 8.7%	χ^2 (3) =2.913, p >.05	
MIPQ Mood abnormally high	2 9.1%	4 4.6%	1 4.5%	2 8.7%	χ^2 (3) =3.887, p >.05	
MIPQ Interest and Pleasure abnormally low	1 4.5%	0 0%	0 0%	0 0%	(χ^2 (3) =3.034, p >.05	
MIPQ Interest and Pleasure abnormally high	2 9.1%	4 19%	0 0%	1 4.3%	(χ^2 (3) =5.878, p >.05	
TAQ Overactivity abnormally high scores	1 4.3%	5 23.8%	0 0%	0 0%	χ^2 (3) =13.403, p< .005	AS>CdLS (p<.05)
TAQ Impulsivity abnormally high scores	3 13%	6 28.6%	1 4.3%	0 0%	χ^2 (3) =10.509, p <.01	AS>CdLS (p<.005)
SCQ at or above cut off for autism spectrum disorder	17 77.3%	12 63.2%	12 60%	15 83.3%	χ^2 (3) =3.484, p >.05	
SCQ at or above cut off for autism	12 54.4%	3 15.8%	4 20%	7 38.9%	χ^2 (3) =8.987, p <.05*	1p36>AS, CdCS (p<.01; .05)

Mood, Interest and Pleasure

The proportion of individuals in each syndrome group scoring abnormally high or low on the MIPQ Mood subscale and the Interest and Pleasure subscale were calculated (the results can be seen in Table 2.6). Fishers exact test analysis of the proportion of individuals showing abnormally high or low affect did not differ significantly across groups (χ^2 (3) =2.913, p >.05 and χ^2 (3) =3.887, p >.05 respectively). Neither were there any significant differences across group on

the proportion of individuals showing abnormally high or low interest and pleasure (χ^2 (3) =5.878, p >.05 and (χ^2 (3) =3.034, p >.05 respectively).

Impulsivity and Over activity

The proportion of individuals in each syndrome group scoring abnormally high on the Activity Questionnaire Impulsivity and Overactivity subscales were calculated (the results can be seen in Table 2.6). Fishers exact test analysis conducted on the proportion of individuals scoring abnormally high on the impulsivity subscale revealed a significant difference across groups (χ^2 (3) =10.509, p <.01). Chi-square post hoc analyses revealed no significant relationships involving the 1p36 syndrome group, although there were significant associations for other groups, as can be seen in Table 2.6.

Fishers exact test analysis conducted on the proportion of individuals scoring abnormally high on the overactivity subscale also revealed a significant difference across groups (χ^2 (3) =13.403, p <.005). Pairwise post hoc Chi-square tests indicated significant differences between groups on overactivity, but none involving the 1p36 group.

SUMMARY

Individuals with 1p36 syndrome have poorer vision than individuals in two of the other syndromes, which is consistent with evidence from medical literature and therefore adds to evidence of poor vision being a phenotypic trait.

On the measures of sociability individuals with 1p36 were found to have lower positive affect (MIPQ) and impulsivity (TAQ) than individuals with Angelman syndrome. The restrictive preferences scale of the RBQ revealed the 1p36 group scored lower than the Cri du Chat group, indicating less restricted preferences for individuals with 1p36. The 1p36 group were more impaired on the SCQ than the Cri du Chat and Angelman syndrome groups, with more

individuals scoring above the cut off for autism, suggesting a higher proportion of autism in 1p36 syndrome than in the two other syndromes.

The results of the comparative analysis between 1p36 syndrome and three other syndromes have been compiled into a single table (Table 2.7) to allow ease of comparison across syndromes and domains. On each domain of the assessments, for each syndrome, the table shows the number of groups each syndrome significantly differs from.

Table 2.7: Relative position of 1p36 syndrome group compared to the other three syndrome groups on assessments of Austism Spectrum Disorder (SCQ), Affect (MIPQ) and hyperactivity (TAQ). (+ = scores higher than one other group, - = scores lower than one other group, o = no difference from other groups).

	Syndrome			
	1p36	AS	CdLS	CdCS
Autism spectrum disorder				
Social Interaction	o	o	+	-
Communication	+	o	o	-
Repetitive Behaviour	o	o	o	o
Affect				
Mood	-	+	o	o
Interest and pleasure	o	o	o	o
Hyperactivity				
Overactivity	o	o	o	o
Impulsivity	-	++	-	o
Repetitive Behaviours				
Stereotyped behaviour	o	o	o	o
Compulsive behaviour	o	o	o	o
Restrictive preferences	-	+	o	+
Repetitive use of language	o	o	o	o
Insistence on Sameness	o	o	o	o

The information in Table 2.7 allows appraisals of performance in more than one domain, across and within syndromes, concurrently with an appraisal of the relative importance of that domain for each syndrome. Whilst further research is required to characterise the syndrome, this comparative approach tentatively suggests a deficit in communication, and reduced affect may all

be part of a social phenotype for 1p36 syndrome. The comparative lack of restricted preferences and impulsivity is probably due to the high frequency of these behaviours in the comparative syndromes.

On the newly developed SQID questionnaire individuals with 1p36 consistently scored higher in familiar interactions compared to unfamiliar, indicating a preference for interaction with known individuals. This result will be further investigated in study two.

STUDY TWO

Whilst study one has added to the research literature on sociability in 1p36 syndrome with the use of standardised assessment measures, the second study provides an observational study of individuals behaviour under various social conditions (high engagement, low engagement and no-engagement). Brief experimental structured social presses were used to evaluate the sociability (social skills and behaviour) displayed by individuals under varied social conditions and with both familiar and unfamiliar adults.

The primary aim of study two is to provide an observational study of the sociability (measured through skills and behaviour) shown by individuals with 1p36 syndrome in experimentally manipulated social settings. It is anticipated that detailed observations, combined with standardised assessment data may provide a basis for a behavioural and specifically a social phenotype for 1p36 syndrome. It is expected, based on data from study one, that higher levels of sociability will be shown with familiar adults compared to unfamiliar (based on data from the SQID) and that higher levels of adult engagement will elicit more social skills and social behaviour in the children.

METHOD

Participants

Recruitment

Of the 25 parents who completed study one, 24 (96%) consented to being contacted regarding study two. Out of the 24 contacted, 19 (79.2%) agreed to participate. However, due to practical limitations such as travel distance, 12 (63.2%) participants were visited.

Participant Information

Twelve participants were included in the study with an age range of 3 years 3 months to 13 years 11 months (mean 6.49, SD 3.56), 8 females (66.7%). Level of ability (self-help skills), mobility

(ability to walk unaided), speech, visual and auditory impairment were described by The Wessex Scale (Kushlick, Blunden & Cox, 1973). Overall 2 (16.7%) were able or partly able, 6 (50%) were mobile, 6 (50%) were verbal (used more than 30 words or signs), 9 (75%) had normal hearing and 3 (25%) had normal vision.

Vineland Adaptive Behavior Scales (Sparrow, Balla, & Cicchetti, 2005) were also completed for each participant and can be seen with individual participant information in Table 2.8. These indicate that all but one child is performing in the significantly low adaptive level relative to peers of the same age.

Table 2.8: Age, gender and scores from the Vineland Adaptive Behavior Scale domains (C=communication, DLS = daily living skills, S=socialization, MS= motor skills) for all participants (P).

P	Gender	Age[1]	C[2]	DLS[2]	S[2]	MS[2*]	Adaptive behaviour composite[2]	Confidence interval	Adaptive level
1	Male	13yrs 11mths	40	47	45	-	43	38-48 (±5)	Low
2	Female	3yrs 3mths	47	46	63	31	44	39-49 (±5)	Low
3	Male	3yrs 2mths	79	64	72	59	65	60-70 (±5)	Low
4	Female	10yrs 11mths	48	50	57	-	50	45-55 (±5)	Low
5	Female	3yrs 3mths	59	62	63	54	55	50-60 (±5)	Low
6	Female	4 yrs 9mths	44	75	72	51	61	57-65 (±4)	Low
7	Female	7yrs 11mths	61	78	61	-	66	62-70 (±4)	Low
8	Female	7yrs 9mths	72	71	73	-	71	67-75 (±4)	Moderately Low
9	Female	11yrs 0mths	42	50	45	-	45	39-54 (±6)	Low
10	Female	4yrs 11mths	47	53	55	54	50	46-54 (±4)	Low
11	Male	4yrs 5mths	67	64	63	56	60	56-64 (±4)	Low
12	Male	11yrs 0mths	56	55	61	-	58	37-49 (±6)	Low

[1] Chronological age in years and months
[2] Standard scores derived from the VABS
*For children age 7years and over the motor skills subscale is not computed

Equipment

Two sets of toys were employed for the social presses, one set for younger or less able participants, and one for older or more able participants (a list of the toys can be found in appendix 1b). A digital video camera was used to record the social presses.

Procedure

Children and parents were visited at a location familiar to the child (either home or school) by two researchers. The social presses were first conducted with an unfamiliar adult (one of the researchers) and then with a familiar adult (either a parent or teacher) in a safe and quiet environment, such as an empty classroom or a room at home. During the social presses both researchers were present, one interacting with the child (unfamiliar condition) and one filming the interactions; or both filming and prompting parents (familiar condition).

The social presses consist of five conditions run consecutively: warm-up/free play (2 minutes), response engagement (3 minutes), non-engagement-1 (3 minutes), active engagement (length of time dictated by completion of activities), and non-engagement-2 (3 minutes). The social presses are run according to a strict protocol (see appendix 1b)

Coding Procedure

The videos of each condition (warm-up, response engagement, non engagement 1, active engagement and non engagement 2) with both familiar and unfamiliar adults were viewed. The child's social behaviour and skills over the course of the whole condition was recorded using a coding schedule (Moss, Yates, Oliver & Howlin, 2009) of fourteen social skills and behaviours(positive affect, negative affect, frequency of physical contact, nature of physical contact, social responsiveness, avoidance of social interaction, social anxiety, spontaneous initiation of interaction, focus of attention, motivation for adult engagement, frequency of eye contact, nature of eye contact, social communication style, quality of social communication skills). The skills and behaviours were coded based on a zero-four scale, with zero typically

indicating lack of a behaviour/skill and four indicating a high presence/intensity (for some behaviours the coding was reversed). Detailed descriptions of the behaviours and coding scales used to code the videos can be found in appendix 1b.

Inter-rater reliability

The data from 7 (58.3%) children were randomly selected for inter-rater reliability of the coding procedure. A second researcher coded all conditions, under the familiar and unfamiliar conditions, for the 7 children using the same procedure as the original coding. Inter-rater reliability was calculated using inter-class correlation, one-way random effects model (judges and participants are treated as random selections, Shrout and Fleiss, 1979).

At item level, this yielded moderate inter-rater reliability for two familiar and one unfamiliar items; substantial reliability for four familiar items and seven unfamiliar; and outstanding reliability for five familiar items and two unfamiliar (Landis & Koch, 1977). Landis & Koch (1977) identified scores falling between 0.40-0.59 as moderate, 0.60-0.79 as substantial and 0.80 and above as outstanding.

The scores for individual behaviours observed across the five conditions were summed into three subscales: skills, motivations and enjoyment. Skills comprises scores from four behaviours: frequency of eye contact, nature of eye contact, social communication style and quality of social communication. Motivation comprises three behaviours: spontaneous initiation of interaction, focus of attention and motivation for adult engagement. Enjoyment comprises seven behaviours: positive affect, negative affect, frequency of physical contact, nature of physical contact, social responsiveness, avoidance of social interaction, and social anxiety. Inter-rater reliability was also calculated at subscale level, which yielded moderate inter-rater reliability for two familiar subscales and three unfamiliar, substantial reliability for three familiar and five unfamiliar, and outstanding reliability for three familiar and two unfamiliar.

Data Analysis

Scores from the fourteen behaviours were summed to create condition scores. For the warm-up and response engagement condition all behaviours except motivation for adult engagement were summed (as the child had constant adult attention). In the active engagement and both non-engagement conditions all behaviours except social responsiveness and avoidance of social interaction were summed (as the adult was not engaging the child). Scores were then analysed for familiarity of adult across conditions.

Kolmogorov-Smirnov tests of normality revealed no violations; therefore the data were subjected to a two-way ANOVA to examine the effect of familiarity and condition on behaviour, followed by t-test post hoc analyses.

RESULTS

A two-way analysis (condition * familiarity) revealed a significant effect of condition ($F_{(4, 40)} = 9.730, p < .001$) but no significant effect of familiarity ($F_{(1, 10)} = 1.871, p > .05$) and no significant interaction ($F_{(4, 10)} = 0.924, p > .05$). Post hoc analysis on the condition effect revealed a number of significant results, as can be seen in Table 2.9.

Table 2.9: Mean (SD's) scores for each condition of the social press with analyses and post hoc comparisons.

Condition					Condition ANOVA		
WU	**RE**	**NE1**	**AE**	**NE2**	**F**	***p***	**Post hoc t-tests**
44.17 (16.16)	42.08 (34.33)	34.33 (15.65)	54.91 (14.22)	33.36 (14.04)	9.730	<.001	AE>WU t = 6.384, df = 10, p <.001 AE>RE t = 5.439, df = 10, p <.001 AE>NE1 t = 6.321, df = 10, p <.001 AE>NE2 t = 4.581, df = 10, p <.001 RE>NE2 t = 1.222, df = 10, p <.001

SUMMARY

The aim of the current study was to examine sociability, through observable social behaviour and social skills, shown by individuals with 1p36 syndrome in experimentally manipulated social settings with both familiar and unfamiliar adults. It was predicted that the sociability shown by individuals would be higher with a familiar than unfamiliar adult, however, the analysis indicated that the children's behaviour and social skills were largely unaffected by the familiarity of the adult.

The analysis of condition indicated that individuals showed more sociability in conditions where the level of engagement from the adult (familiar and unfamiliar) was high. The active engagement condition seemed to evoke increased social behaviour and skills more than the other high level condition (warm-up), which could be due to the level of active direction the adult provides for the child in this condition.

GENERAL DISCUSSION

The aim of the current research was to characterise the behavioural, and in particular the social, phenotype for Monosomy 1p36 deletion syndrome. At present the syndrome has received a lot of interest regarding the genetic and clinical features and implications of the condition; however, research has not focused on the behaviour and social characteristics of individuals with 1p36 syndrome. This study is the first to concentrate on the sociability (defined as the relative tendency or disposition to be sociable or associate with other people and measured through a number of social constructs) of individuals with 1p36 deletion syndrome. It is also the first to use psychometrically robust measures of sociability with individuals with Monosomy 1p36, to use comparison groups of individuals with other genetic syndromes matched on level of ability and to use direct observation methods which allow an investigation of sociability under different experimental conditions.

In study one a number of standardised measures of aspects of sociability were completed by parents and carers of individuals with 1p36 syndrome. The findings from these questionnaires were compared to information from comparable individuals with other genetic syndromes (Angelman syndrome (AS), Cornelia de Lange syndrome (CdLS) and Cri du Chat syndrome (CdCS)). This revealed that individuals with 1p36 have poorer vision than individuals in two of the comparative syndromes. Visual impairments are well documented in the condition suggesting they are phenotypic (Battaglia et al., 2008; Gajecka et al., 2007). The comparisons also revealed that the 1p36 group scored lower than the AS group on mood and impulsivity. This is in agreement with a recent study (Oliver et al., 2009) which found that AS is characterised by elevated positive affect and hyperactivity. It has also been found that low mood is positively correlated with health problems (Berg, Arron, Burbidge, Moss & Oliver, 2007) and given the high incidence of health problems found in 1p36 it is possible that this offers an explanation for the lowered affect seen in the group (MIPQ). Parents particularly reported problems with reflux, which has been found to have an association with low mood in Cornelia de Lange syndrome (Luzanni et al., 2003) suggesting another possibility for the observed low mood in children with 1p36.

It was also found that individuals with 1p36 syndrome scored higher than individuals with CdCS on communication, indicating more impairment. This could have been one of the reasons that a higher proportion of individuals with 1p36 scored above the cut off for autism compared to those with CdCS. Although a recent study (Oliver et al., 2009) did not find a predisposition to autism in CdCS, therefore the conclusion that individuals with 1p36 syndrome show elevated levels of autism should be treated with caution. Whilst autistic features have been reported in 1p36 syndrome (Slavotinek et al., 1999; Battaglia, 2005; Knight-Jones et al., 2005) there has been no detailed description of the features considered to be typical of autism and no standardised assessments of autism have been reported. Furthermore, on the Repetitive Behaviour

Questionnaire individuals with CdCS showed more restricted preferences compared to individuals with 1p36 syndrome, although this is a well documented finding in individuals with CdCS and can therefore not be taken as evidence that individuals with 1p36 do not show signs of this behaviour. The findings regarding potential indicators of a prevalence of autism in 1p36 syndrome are therefore inconclusive.

Using a relatively new measure, the Sociability Questionnaire for Intellectual Disabilities (SQID; Collis & Oliver, 2007) suggests that individuals with 1p36 syndrome are more sociable (defined through aspects of social interaction) with familiar people. The suggested clinical cut off's for the questionnaire also suggest that when individuals are with familiar people they are more likely to show signs of excessive sociability, and when with unfamiliar people show signs of excessive shyness. However, this questionnaire has yet to be verified and no comparison data exist for this measure, making conclusions difficult.

In study two the possibility that individuals are more sociable with familiar adults was assessed in a structured experimental paradigm (a social press) as well as investigating the impact that varying levels of adult engagement might have on the children's sociability (measured through observable social skills and behaviour). Based on information from parents and carers it was predicted that individuals would show higher levels of sociability with familiar adults compared to unfamiliar. It was also predicted that sociability would be higher in conditions where the adult (familiar and unfamiliar) showed the child high levels of attention/engagement compared to conditions where engagement was only given as a response to the child initiating it, or not given at all.

It was found that overall children did not show higher levels of sociability with familiar adults. It had been predicted that the children would be more sociable with familiar adults as there had been a significant difference between parents' ratings of their child's sociability with familiar and unfamiliar people on the SQID. However, one potential reason for the difference between the social presses and the SQID data is the context of the social encounters. Although the unfamiliar

adult social presses were conducted first, they were conducted in a safe and familiar environment to the child. Typically the child had been introduced to the unfamiliar adult by a teacher (a trusted adult) and the children had been told by parents that someone would be visiting them; therefore some of the anxiety at being around an unfamiliar person might have been alleviated.

The prediction that individuals would show more sociability in conditions where the adult's level of attention to the child was greatest has been supported. For both familiar and unfamiliar adults the active engagement condition yielded the highest scores. In this condition the children are encouraged to join in the interaction with the adult using novel toys and games and the adult continually gives the child high levels of engagement. It can be seen that in such a condition children who do not have social deficits (based on data from study one) would show high levels of sociability. It was also found that in the non-engagement conditions there were consistently low levels of sociability shown by the children.

In study two Vineland Adaptive Behavior Scales were also completed for all participants, which indicated that the group were performing at comparatively low levels compared to age matched peers. The profile scores across the domains of the Vineland also indicate that the majority of the children obtained higher scores on the socialization domain compared to the communication and daily living skills domains. In clinical trials of the Vineland, it has been found that individuals with Autism score lower in the socialization domain compared to the other two. The profile obtained in the current study is more indicative of a generalised intellectual disability (Sparrow, Cicchetti & Balla, 2005). On the communication subscale of the Vineland individuals were typically more than two standard deviations below the mean, again indicating significantly low performance compared to peers, which complies with the data from the social communication questionnaire in study one.

Overall the comparative approach suggests a deficit in communication (indicative of a delay in communication development as measured by the SCQ, Vineland and Wessex questionnaires and

not indicative of an autistic spectrum communication deficit) and reduced affect, may be part of a social phenotype for 1p36 syndrome. The SQID data suggest that individuals may be more sociable with familiar adults, possibly showing excessive sociability when around familiar people. The social press observations suggest that the under conditions of high engagement with an adult the children show higher levels of sociability than low engagement conditions, with both familiar and unfamiliar people.

Limitations of the Research

As with any research there are areas for improvement with the current study and limitations of what could be achieved. One of the main considerations is that of participant selection. Participants were recruited via a support group and it is possible that the group attracts families who are finding it difficult to cope with their child's condition and/or who have limited external resources. Typically in the published research on 1p36 syndrome participants have been recruited via genetics departments at large teaching hospitals, therefore possibly giving a wider range of participants compared to a self-selecting support group sample.

The comparison groups employed for the current study were chosen based on their comparative level of ability on the Wessex questionnaire and as such formed good comparative matched participants for the 1p36 sample. However, there are known characteristics of some of the syndromes which have been highlighted by the current research. For example, it is well known that individuals with Angelman syndrome have excessive impulsivity and individuals with Cri du Chat syndrome show excessive preference for individual objects. Both of these characteristics were found in the comparative study in the current research, and compared to these two syndromes individuals with 1p36 would appear to show a lack of impulsivity and lack of restricted preferences. However, when viewed in comparison to the excesses in both of these traits exhibited by AS and CdCS the results are not surprising and therefore should not be taken

as indicative of a social phenotype for 1p36 syndrome. It would therefore be advantageous for future research to investigate differences and similarities with other syndrome groups.

The Social Communication Questionnaire is also recommended for use with individuals with a mental age above 2-years (Rutter, Bailey & Lord, 2003). However, recent research (Lee, David, Rusyniak, Landa & Newschaffer, 2007) does indicate its efficacy when used with younger, less able individuals; therefore the results can be considered valid. It should be remembered, however, that on the Wessex questionnaire only 50% of the children were reported to be verbal (having more than 30 words or signs) which could impact on any results of communication measures. Although the results from the communication subscale of the SCQ were validated by the scores on the Vineland scale also.

The lack of comparison data and psychometric information on the sociability questionnaire for intellectual disabilities (SQID) means that at this time conclusions cannot be extracted from the questionnaire without due caution. There is a similar difficulty with the use of the social presses. Although good inter-rater reliability was obtained on the coding of the presses, this is the first study utilising the method and again, conclusions must be drawn with caution.

The use of parents as a familiar adult may pose some difficulties with the social presses. Although very clear instructions are given to parents and constant prompts are provided throughout the social presses, many parents found it extremely difficult to ignore their child in the non-engagement and response engagement conditions. Some parents freely initiated interaction even when directed by the researchers to refrain from doing this. Whilst it is acknowledged that it must be very difficultly and unnatural for parents to ignore their children when they are in such close proximity, it is a limitation which should be addressed in future research.

Clinical Implications and Future Directions

There is a high prevalence of autism reported in many genetic syndromes and it has been reported in six studies on 1p36 syndrome . However, there have been no standardised assessments of autism reported. In the present research proportionally more of the 1p36 group scored above the cut off for autism that the Cri du Chat group. However, given there is not a predisposition to autism in Cri du Chat syndrome the results do not provide evidence of a high incidence of autism in the 1p36 group. Research needs to be conducted into the specific features of autism reported in the literature, such as reduced social interaction, repetitive stereotypies, tendency to beat, smell or roll objects repetitively, manual apraxia and poor eye contact (Blennow et al., 1995; Tiong et al., 2005; Battaglia et al., 2008) as no evidence of these difficulties was found in the current research. The implications of a diagnosis of autism are far reaching, not just for the individual and their carer but also for clinical services.

Evidence from the social presses that individuals enjoy high engagement could be harnessed when interventions are necessary for problems such as temper tantrums or self-injury as the children appear to find social interaction enjoyable. Clinicians should also be aware of the potential for individuals to be sociable even with unfamiliar adults, and perhaps build such knowledge into interventions e.g. social skills training.

It is important for clinicians working with children and families affected by 1p36 syndrome to be aware of the behavioural and social characteristics of the syndrome and not just the genetic and molecular features.

Summary

The aim of the current research was to begin to delineate the behavioural and social phenotype of 1p36 deletion syndrome. The current study provides evidence that tentatively suggests 1p36 may be characterised by a delay in communication development and therefore a deficit in this social

skill, reduced affect, higher sociability with familiar adults under certain conditions, and higher sociability under conditions of high attention/engagement.

However, given that this is the first study to begin to describe and compare 1p36 syndrome, it is difficult and unwise to make generalised conclusions about the sociability of individuals with the condition. At the present time it would appear that there is not a high prevalence of autism in the syndrome (as measured by the SCQ and RBQ), sociability does not appear excessive (as measured by the SQID and social presses) and social skills do not appear particularly impaired (measured by the SCQ, RBQ, SQID and social presses).

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APPENDICES VOLUME I

Appendix 1a: Information and letters to parents
Consent forms
Questionnaire pack
Confirmation of ethical approval

Appendix 1b: List of toys employed in the social presses
Instructions for running the social presses
Behaviour descriptions and coding schedule

Appendix 2: Public domain briefing document

Appendix 3: Instructions for authors and notes for submission for journals

Appendix 1a

Information and letters to parents

Consent forms

Questionnaire pack

Confirmation of ethical approval

Unique RARE CHROMOSOME DISORDER SUPPORT GROUP ...but not alone

UNIVERSITY OF BIRMINGHAM

UNIVERSITY OF BIRMINGHAM RESEARCH INTO CHARACTERISTICS OF INDIVIDUALS WITH MONOSOMY 1P36 DELETION SYNDROME

This booklet should contain:

1. Letter of invitation
2. Professor Chris Oliver's contact details (See letter of invitation)
3. Information sheet
4. Consent forms
5. Questionnaires

Instructions for Completing Questionnaires:

1. The questionnaires should be completed by the **main caregiver.**

2. When you have completed the questionnaires, please check that you have answered every question, and return them to us in the *freepost* envelope provided.

Thank you for agreeing to participate in this research.

RARE CHROMOSOME DISORDER SUPPORT GROUP
...but not alone

Edgbaston
Birmingham
B15 2TT

Project Director: Professor
Chris Oliver
Tel: 0121 414 7206

Date

Dear Parent or Carer,

We would like to invite you and the person you care for to take part in a new research project being carried out at the University of Birmingham in partnership with UNIQUE The research has the full support of UNIQUE and a good response will provide valuable information for the group. Briefly, the research is a questionnaire study looking at different behaviours in children and adults with Monosomy 1p36 Deletion Syndrome.

There is an information sheet enclosed that gives you more details about why the research is being carried out and what it will involve. If you feel it is appropriate you may wish to discuss the research with the person you care for before a decision is made about taking part.

Please read the information sheet before completing a consent form and the questionnaires and if you are unclear about any aspect of the study or have any questions then contact Professor Chris Oliver at the above address or on 0121 414 7206.

When we have completed the study we will send you a personalised feedback report with information about the person you care for and a report will be written for the newsletter.

If you wish to take part please complete a consent form and the questionnaires and return them to us in the envelope provided. Thank you for your time and we look forward to hearing from you.

Yours sincerely,

Signature of chair		
Name of chair	837009	Chris Oliver
Information officer	DClinPsy	Professor of Clinical Psychology
UNIQUE		

INFORMATION SHEET

Background:
A team at the University of Birmingham is carrying out a study to look at several aspects of behaviour in people with Monosomy 1p36 Deletion Syndrome.

As a research team we would like to investigate the specific behavioural and developmental characteristics of Monosomy 1p36 Deletion Syndrome. This would allow us to describe the behavioural phenotype for the syndrome-that is, the specific and characteristic behavioural repertoire exhibited by individuals with a genetic or chromosomal disorder. Behavioural phenotypes are important as they lead to a greater understanding of behaviour in rare syndromes. This can then lead to an increase in awareness of the potential problems which might arise and in the development and provision of early interventions for these problems.

We would also like to investigate some specific problems which have been reported for individuals with Monosomy 1p36 Deletion Syndrome. These are problems with over-eating and self injurious behaviour. Both of these characteristics can be distressing for parents and carers as well as professionals, and we would like to gain more insight into these behaviours. This would be done by observing individuals in a range of different situations.

Further, there are many reports of cognitive impairments for individuals with Monosomy 1p36 Deletion Syndrome; however, the literature is not very helpful in telling us exactly what the impairments are. We would therefore like to conduct some simple cognitive assessments with some individuals to try to establish what impairments are present and how severe these are.

What does it involve?
The questionnaire pack you have received contains several short questionnaires about some aspects of behaviour. The questionnaire pack should take no longer than 30 minutes to complete.

You will also be invited along to Birmingham University for the day to meet other families with individuals with Monosomy 1p36 Deletion Syndrome. We will be meeting with parents/carers for no more than one hour each to discuss in more detail the person they care for with Monosomy 1p36 Deletion Syndrome.

In addition, we are looking into feeding/eating difficulties and self-injury in individuals with Monosomy 1p36 Deletion Syndrome. We would like to visit individuals (and their carers) at their home/day placement/school etc to carry out some observations in different situations.

We are also researching the specific cognitive problems individuals with Monosomy 1p36 Deletion Syndrome have. We would like to either visit individuals at their home/day placement/school etc or invite individuals and carers along to the research centre at the university to carry out some simple cognitive assessment. These will be able to give us information on the specific problems which may be faced by individuals with Monosomy 1p36 Deletion Syndrome, for example, memory, attention, concentration problems etc.

What are the benefits/drawbacks of taking part?

Whilst we cannot promise any direct benefits to you for agreeing to take part in the study, the information that we gather will be invaluable in increasing the understanding of Monosomy 1p36 Deletion Syndrome. We hope that a greater understanding of the cognitive impairments and behavioural characteristics of the syndrome would lead to the development of appropriate interventions for these problems.

We do not anticipate any drawbacks of taking part in this study; in fact we hope that you would find it a very worthwhile experience. However, it is important to note that the interview about experiences of receiving a diagnosis may prompt emotional memories of a difficult time, which may cause you to feel distressed. If at any point you feel distressed then it is possible to pause or terminate the interview. The experience of discussing potentially distressing events can have a positive effect on participants as it may be an opportunity for participants to voice their experiences; something which they may not have had the opportunity to do before.

Consent:
It is up to you whether or not you want your child or the person you care for to take part in the study. If your child / the person you care for is under the age of 16 or over the age of 16 but unable to give consent then please fill in consent form A on their behalf. If your child or the person you care for is over the age of 16 and is able to give consent for themselves, please ask them to complete consent form B. If you feel that it is appropriate, you may wish to discuss the project with your child or the person you care for.

If you do not wish to take part in all aspects of the research, then there is the opportunity to take part in only the questionnaire study, if you so wish.

Withdrawal:
Should you or the person you care for decide that you no longer wish to be involved in the study, the information that you have provided can be withdrawn at any time without you giving a reason. Even after the questionnaire, interview and observations have been completed, consent can be withdrawn and any data collected will be destroyed. This will not restrict access to other services and will not affect the right to treatment.

Confidentiality:
Contact has been made through your syndrome support group and we do not know any of your personal details at this stage. All details collected will be kept on a confidential database that is only accessible to those working on the project. Anonymity is ensured by storing the questionnaire data separately from any material that identifies participants. If published, information will be presented without reference to any identifying information.

At the end of the study:
Each parent/ carer will receive a personalised feedback report on their child or the person they care for. A summary of the project's findings will be circulated to anyone involved who wishes to see a copy and a report will be written for the newsletter. Any requests for advice concerning your child/the person you care for will be referred to Professor Chris Oliver, Clinical Psychologist. It is possible that you may be invited to participate in further research after the study however, consenting to participate in this study does not mean that you are obliged to do so.

Review:
This study has been reviewed by the University of Birmingham, School of Psychology Research Ethics Committee. If you have any concerns about the conduct of this study please contact Professor Orford at the School of Psychology Ethics Committee, University of Birmingham, Edgbaston, Birmingham, B15 2TT.

Thank you very much for taking the time to read this information.

Consent Forms

Behaviour in Monosomy 1p36 Deletion Syndrome

CONSENT FORM A: For people aged below 16 or people over 16 who are unable to give consent.

Please initial the boxes

I confirm that I have read and understood the information sheet for the above study and have had the opportunity to ask questions. ☐

I understand that participation in the study is voluntary and that I am free to end my child's/the person I care for's involvement at any time, or request that the data collected in the study be destroyed, without giving a reason. ☐

I agree to the participation of my child's / the person I care for in the above study ☐

Please complete the information below

Participant's name…….date of birth……

Parent or guardian's name……Mr/Mrs/Miss/Ms (please circle)

Parent or guardian's signature……Date……..

Please state relationship with participant…….

FOR OFFICE USE ONLY

Signature of researcher……..Date……...

Behaviour in Monosomy 1p36 Deletion Syndrome

CONSENT FORM B: For people aged 16 or over who are able to give consent.

Please initial the boxes

I confirm that I have read and understood the information sheet for the above study or that it has been explained to me and have had the opportunity to ask questions. ☐

I understand that participation in the study is voluntary and that I am free to end my involvement at any time, or request that the data collected in the study be destroyed, without giving a reason. ☐

I agree to take part in the above study ☐

Please complete the information below

Your name……. Your date of birth……

Your signature……Date…….

FOR OFFICE USE ONLY

Signature of researcher……..Date……...

THE UNIVERSITY
OF BIRMINGHAM

The Centre for Neurodevelopmental Disorders
School of Psychology
University of Birmingham

RESEARCH INTO CHARACTERISTICS OF INDIVIDUALS WITH MONOSOMY 1P36 DELETION SYNDROME

Instructions for Completing Questionnaire:

1. **The questionnaires should be completed by the <u>main caregiver.</u>**

2. **When you have completed the questionnaire, please check that you have answered every question, and return them to us at the following address:**

THE UNIVERSITY
OF BIRMINGHAM

The Centre for Neurodevelopmental Disorders
School of Psychology
University of Birmingham

RESEARCH INTO CHARACTERISTICS OF INDIVIDUALS WITH MONOSOMY 1P36 DELETION SYNDROME

Questionnaires included in this pack are:

BACKGROUND INFORMATION
SOCIAL COMMUNICATION QUESTIONNAIRE: LIFETIME
THE ACTIVITY QUESTIONNAIRE
THE GRQ
THE SOCIABILITY QUESTIONNAIRE FOR PEOPLE WITH INTELLECTUAL DISABILITIES
HEALTH QUESTIONNAIRE
WESSEX QUESTIONNAIRE
REPETITIVE BEHAVIOUR QUESTIONNAIRE
THE MOOD, INTEREST AND PLEASURE QUESTIONNAIRE (SHORT FORM)
THE CHALLENGING BEHAVIOUR QUESTIONNAIRE
THE FOOD-RELATED PROBLEMS QUESTIONNAIRE

BACKGROUND INFORMATION

Please tick or write your response to these questions concerning background details:

1. Today's date: _____

2. Your name:

_____

Your address:

_____

_____

Your phone number:

_____

3. Would you be happy to be contacted for future research? Yes ☐ No ☐

*The following questions regard information about the **person you care for**:*

1. Name of person: _____ Gender: Male ☐ Female ☐

2. Date of Birth: ___/___/____ **Age:**_____

3. Is the person verbal? (i.e. speaks / signs more than 30 words) Yes ☐ No ☐

4. Is the person able to walk unaided? Yes ☐ No ☐

5. Has the person been diagnosed with a syndrome? Yes ☐ No ☐

If yes, please answer the rest of this questionnaire. If no, please move on to question 10.

6. Which syndrome has the person been diagnosed with?

_____

7. When was the person diagnosed? _____

8. Who diagnosed the person?

Paediatrician ☐ Clinical Geneticist ☐

GP ☐ Other

_____

9. If the person has had a blood test to determine the cause of their genetic syndrome, please answer the rest of question 9. If not, please move on to question 10.

9a. When was the blood test carried out? _____

9b. Where was the blood test carried out? _____

9c. Who carried out the blood test? _____

9d. Can we contact the person to request the test results? Yes ☐ No ☐

If yes, please sign to provide consent _____

10. Has the person experienced any of the following life events in the *past twelve months*:-

Yes No N/A

10a. Significant change of staff or friends at residential unit? ……...

10b. Significant change of staff or friends at day provision? …….....

10c. Significant change in day provision, e.g. school, college or job placement? ……

10d. Significant change in place of residence? ……..

10e. Serious illness and / or hospitalisation? ……..……

10f. Serious illness of a close relative, close friend or close member of staff? …….

10g. Death of a close relative, close friend or close member of staff? ……...

10h. Parents divorced or separated? ……...

Other (please give details) _____

_____

11a. Has the person ever suffered from gastro-oesophageal reflux since they were 3 years old? *(please circle)*

Yes (I'm certain they have)	No (I'm certain they have not)	I don't know

11b. Does the person suffer from gastro-oesophageal reflux now? *(please circle)*

Yes (I'm certain they do)	No (I'm certain they do not)	I don't know

12a. When was the person's gastro-oesophageal reflux first diagnosed (if applicable)?

12b. When did the person last see a Gastro-intestinal specialist regarding gastro-oesophageal reflux?

(please circle)

Within the last month	Within the last 6 months	Within the last year	Over a year ago	Never

12c. Has the person had a surgical procedure to treat gastro-oesophageal reflux, e.g. Nissan fundoplication? Yes ☐ No ☐

If yes, **what** was the surgical procedure and **when** was it done?

12d. Is the person currently taking any medication for gastro-oesophageal reflux?

Yes ☐ No ☐

If yes, please list any **medication** taken for reflux, the **dosage** of the medication and **when** it is taken?

12e. Has the person had any other treatments for gastro-oesophageal reflux?

Yes ☐ No ☐

If yes, please state **what** treatments have been used and **when** they were used?

13. Has the person ever completed an intelligence test given by a Psychologist or other professional?

Yes ☐ No ☐

If 'Yes' please tick the I.Q. score given if known:

Superior ☐ Moderate ☐ Average ☐ Severe ☐

Borderline ☐ Profound ☐ Mild ☐ Do not know ☐

14. Does the person attend:

(please tick box and underline any words that apply)

Mainstream school	☐	Adult Day Centre	☐
Special school (mild, moderate, severe)	☐	Other Facility (please specify)	☐
College (residential / adults with disabilities / mainstream)	☐		

15. Are you the person's: (please tick appropriate box)

Father ☐ Mother ☐ Other ☐

If other please specify relationship_____

Thank you, please check your answers and go on to the first questionnaire.

SOCIAL COMMUNICATION QUESTIONNAIRE (SCQ): LIFETIME

The following questions relate to the person you care for. Please answer each question by circling *yes* or *no*. A few questions ask about several related types of behaviour; please circle *yes* if *any* of these behaviours have ever been present. Although you may be uncertain about whether some behaviours were ever present or not, please answer *yes* or *no* to every question on the basis of what you think.

1. Is she/he now able to talk using short phrases or sentences? ……yes no

If *No*, skip to question 8.

2. Can you have a to and fro "conversation" with her/him that involves taking turns or building on what you have said? ……yes no

3. Has she/he ever used odd phrases or said the same thing over and over in almost exactly the same way (either phrases that she/he has heard other people use or ones that she/he has made up? ……yes no

4. Has she/he ever used socially inappropriate questions or statements? For example, has she/he ever regularly asked personal questions or made personal comments at awkward times? ……yes no

5. Has she/he ever got her/his pronouns mixed up (e.g., saying *you* or *she/he* for *I*)? …...yes no

6. Has she/he ever used words that she/he seemed to have invented or made up her/himself; put things in odd, indirect ways; or used metaphorical ways of saying things (e.g., saying *hot rain* for *steam*)? ……yes no

7. Has she/he ever said the same thing over and over in exactly the same way or insisted that you say the same thing over and over again? ……yes no

8. Has she/he ever had things that she/he seemed to have to do in a very particular way or order or rituals that she/he insisted that you go through? …….yes no

9. Has her/his facial expression usually seemed appropriate to the particular situation, as far as you could tell? ……..……yes no

10. Has she/he ever used your hand like a tool or as if it were part of her/his own body (e.g., pointing with your finger, putting your hand on a doorknob to get you to open the door)? ……yes no

11. Has she/he ever had any interests that preoccupy her/him and might seem odd to other people (e.g., traffic lights, drainpipes, or timetables)? ……..…….yes no

12. Has she/he ever seemed to be more interested in parts of a toy or an object (e.g., spinning the wheels of a car), rather than using the object as it was intended? ……yes no

13. Has she/he ever had any special interests that were *unusual* in their intensity but otherwise appropriate for her/his age and peer group (e.g., trains, dinosaurs)? …...yes no

14. Has she/he ever seemed to be *unusually* interested in the sight, feel, sound, taste, or smell of things or people? ……. yes no

15. Has she/he ever had any mannerisms or odd ways of moving her/his hands or fingers, such as flapping or moving her/his fingers in front of her/his eyes? ….....yes no

16. Has she/he ever had any complicated movements of her/his whole body, such as spinning or repeatedly bouncing up and down? ……yes no

17. Has she/he ever injured her/himself deliberately, such as by biting her/his arm or banging her/his head? ……yes no

18. Has she/he ever had any objects (*other* than a soft toy or comfort blanket) that she/he *had* to carry around? …….yes no

19. Does she/he have any particular friends or a best friend? ……yes no

20a. Have you known the person since they were 4 years old? ……..…yes no

For the following questions, please focus on the time period between the person's fourth and fifth birthdays. You may find it easier to remember how things were at that time by focusing on key events, such as starting school, moving house, Christmas time, or other specific events that are particularly memorable for you as a family. If your child is not yet 4 years old, please consider her or his behaviour in the past 12 months.

If you have not known the person since they were 4 years old, please leave questions 20 to 40 blank and go on to the next questionnaire.

20. When she/he was 4 to 5, did she/he ever talk with you just to be friendly (rather than to get something)? ……..…..yes no

21. When she/he was 4 to 5, did she/he ever *spontaneously* copy you (or other people) or what you were doing (such as vacuuming, gardening, or mending things)? ……yes no

22. When she/he was 4 to 5, did she/he ever *spontaneously* point at things around her/him just to show you things (not because she/he wanted them)? ……..…….yes no

23. When she/he was 4 to 5, did she/he ever use gestures, other than pointing or pulling your hand, to let you know what she/he wanted? ……..……yes no

24. When she/he was 4 to 5, did she/he nod her/his head to mean *yes*? ……..……yes no

25. When she/he was 4 to 5, did she/he shake her/his head to mean *no*? ……..……..….yes no

26. When she/he was 4 to 5, did she/he usually look at you directly in the face when doing things with you or talking with you? ……..……..…yes no

27. When she/he was 4 to 5, did she/he smile back if someone smiled at her/him? ….yes no

28. When she/he was 4 to 5, did she/he ever show you things that interested her/him to engage your attention? ……..…yes no

29. When she/he was 4 to 5, did she/he ever offer to share things other than food with you? ……..……yes no

30. When she/he was 4 to 5, did she/he ever seem to want you to join in her/his enjoyment of something? ……..…..…yes no

31. When she/he was 4 to 5, did she/he ever try to comfort you if you were sad or hurt? ……....yes no

32. When she/he was 4 to 5, when she/he wanted something or wanted help, did she/he look at you and use gestures with sounds or words to get your attention? ……..……yes no

33. When she/he was 4 to 5, did she/he show a normal range of facial expressions? ……yes no

34. When she/he was 4 to 5, did she/he ever spontaneously join in and try to copy the actions in social games, such as *The Mulberry Bush* or *London Bridge Is Falling Down*? ……..... yes no

35. When she/he was 4 to 5, did she/he play any pretend or make-believe games? .... yes no

36. When she/he was 4 to 5, did she/he seem interested in other children of approximately the same age whom she/he did not know? ……...…. yes no

37. When she/he was 4 to 5, did she/he respond positively when another child approached her/him? ……...yes no

38. When she/he was 4 to 5, if you came into a room and started talking to her/him without calling her/his name, did she/he usually look up and pay attention to you? …… yes no

39. When she/he was 4 to 5, did she/he ever play imaginative games with another child in such a way that you could tell that they each understood what the other was pretending? ……..…..….yes no

40. When she/he was 4 to 5, did she/he play cooperatively in games that required joining in with a group of other children, such as hide-and-seek or ball games? ……...yes no

Thank you, please check your answers and go on to the next questionnaire.

THE ACTIVITY QUESTIONNAIRE

Instructions:

- Please read each item carefully and circle the appropriate number on the scale, for the person you care for.
- Please ensure that you indicate a response for every item. If the particular behaviour does not apply,
  for example, if the person is not verbal or not mobile, please circle 0 on the scale.

		Never/ almost never	Some of the time	Half of the time	A lot of the time	Always/ almost all the time
1.	**Does the person wriggle or squirm about when seated or lying down?**	**0**	**1**	**2**	**3**	**4**
2.	Does the person fidget or play with their hands and/or feet when seated or lying down?	**0**	**1**	**2**	**3**	**4**
3.	**Does the person find it difficult holding still?**	**0**	**1**	**2**	**3**	**4**
4.	**Does the person find it difficult to remain in their seat even when in situations where it would be expected?**	**0**	**1**	**2**	**3**	**4**
5.	**Does the person prefer to be moving around or becomes**	**0**	**1**	**2**	**3**	**4**
6.	**When the person is involved in a leisure activity (e.g. watching TV, playing a game etc.) do they make a lot of noise?**	**0**	**1**	**2**	**3**	**4**
7.	**When the person is involved in an activity, are they boisterous and/or rough?**	**0**	**1**	**2**	**3**	**4**
8.	**Does the person act as if they are "driven by a motor" (i.e. often very active)?**	**0**	**1**	**2**	**3**	**4**
9.	**Does the person seem like they need very little rest to recharge their battery?**	**0**	**1**	**2**	**3**	**4**
10.	**Does the person often talk excessively?**	**0**	**1**	**2**	**3**	**4**
11.	**Does the person's behaviour seem difficult to manage/contain whilst out and about (e.g. in town, in supermarkets etc.)?**	**0**	**1**	**2**	**3**	**4**
12.	**Do you feel that you need to "keep an eye" on the person at all times?**	**0**	**1**	**2**	**3**	**4**
13.	**Does the person you care for seem to act/do things without stopping to think first?**	**0**	**1**	**2**	**3**	**4**
14.	**Does the person blurt out answers before questions have been completed?**	**0**	**1**	**2**	**3**	**4**
15.	**Does the person start to respond to instructions before they have been fully given or without seeming to understand them?**	**0**	**1**	**2**	**3**	**4**
16.	**Does the person want things immediately?**	**0**	**1**	**2**	**3**	**4**
17.	**Does the person find it difficult to wait?**	**0**	**1**	**2**	**3**	**4**
18.	**Does the person disturb others because they have difficulty waiting for things or waiting their turn?**	**0**	**1**	**2**	**3**	**4**

Thank you, please check your answers and go on to the next questionnaire.

The GRQ

Instructions:

- This questionnaire asks about behaviours sometimes shown by people with learning disabilities.
- Please read the questions and examples carefully and indicate how often each behaviour has occurred in the **last two weeks** by circling the appropriate answer.

Does the person you care for:	**More than once an hour**	**Once an hour**	**Once a day**	**Once a week**	**Not occurred**
1. Arch his/her back, lie over arms of chairs or people on his/her back? ……	4	**3**	**2**	**1**	**0**
2. Lie over an object on his/her stomach? e.g. a side of an arm chair. ……. ..	4	**3**	**2**	**1**	**0**
3. Salivate excessively? …….	4	**3**	**2**	**1**	**0**
4. Fidget, wriggle or move their body a great deal? ……	4	**3**	**2**	**1**	**0**
5. Place their hands or fingers in back of their mouth? ……...	4	**3**	**2**	**1**	**0**
6. Chew on his/her clothes, fingers, hands or other parts of the body, objects or material? ……..	4	**3**	**2**	**1**	**0**
7. Grind their teeth? …….	4	**3**	**2**	**1**	**0**
8. Scratch, hit, press or rub around the upper chest or throat? ……	4	**3**	**2**	**1**	**0**
9. Drink, request or seek out an excessive amount of fluids? …….	4	**3**	**2**	**1**	**0**
10. Cough, gag or regurgitate? ……	4	**3**	**2**	**1**	**0**
11. Appear in pain or discomfort (cry, groan or moan)? …….	4	**3**	**2**	**1**	**0**
12. Refuse food even though they are probably hungry? ……	4	**3**	**2**	**1**	**0**

13. Does the person you care for appear indecisive about food (edging towards table or food then moving away

repeatedly, taking food and putting it back)? *(please tick)* **Yes** ☐ **No** ☐

14. Does the person you care for wake during the night?	**Never**	**Once a week**	**Most nights**	**Every night**
15. Does the person you care for sleep sitting or propped up?	**Never**	**Once a week**	**Most nights**	**Every night**
16. Does the person you care for seem to have bad breath?	**Never**	**Once a week**	**At the same time everyday**	**All day every day**

17. Has the person you care for prone to respiratory tract infections? *(please tick)* **Yes** ☐ **No** ☐

If 'yes' please indicate how often they occur:	**Monthly**	**Quarterly**	**Every six months**	**Annually**

Other (please specify)_____

Thank you, please check your answers and go on to the next questionnaire.

THE SOCIABILITY QUESTIONNAIRE FOR PEOPLE WITH INTELLECTUAL DISABILITIES (SQID)

Instructions:

This questionnaire asks you how the person you care for typically behaved in social situations over the last two months. Each situation will involve one of the following:

1. *The person's main caregiver*: Someone that provides the main support and care for the person, e.g. a parent or carer.

2. *A familiar adult or someone familiar of the same age*: Someone that knows the person relatively well but does *not* provide the main care for the person, e.g. a relative *not* in the immediate family, a friend of the family, a support worker at school / college, a friend at school / college etc.

3. *An adult or someone of the same age that the person does not know*: Someone the person has *never* met before, e.g. a stranger, a new teacher, a new support worker at school / college, someone new of the same age at school / college etc.

The person may appear 'sociable', 'shy' or somewhere in between in the situations given below.

- If the person is 'sociable' (s)he may show one or more of the following behaviours: looks pleased; starts to speak or sign to others; turns face and / or body towards others; or tries to gain other people's attention in someway.

- If the person is 'shy' (s)he may show one or more of the following behaviours: looks a little sad or distressed; reluctant to speak or sign to others; turns head and / or body away from others; tries to avoid or remove himself / herself from situations when other people are present.

Read each question and circle the response that best describes the behaviour of the person in the situation described.

For example, for question 4 if you think that when the person is spending time with a familiar adult (s)he would be 'very sociable' then your answer would look like this:-

4. (S)he is spending time with a familiar adult? 1 2 3 4 5 6 (7)

How would the person you care for appear if…	**Very shy**	**Moderately shy**	**A little shy**	**Neither**	**A little sociable**	**Moderately sociable**	**Very sociable**
1. Her / his main caregiver walks up to her / him?	**1**	**2**	**3**	**4**	**5**	**6**	**7**
2. (S)he is spending time with an adult (s)he does *not* know? ……...	**1**	**2**	**3**	**4**	**5**	**6**	**7**
3. Someone (s)he does *not* know that is her / his own age walks up to her /him? …….	**1**	**2**	**3**	**4**	**5**	**6**	**7**
4. (S)he is spending time with a familiar adult? …….	**1**	**2**	**3**	**4**	**5**	**6**	**7**
5. (S)he is the focus of attention in a group of adults (s)he knows? ……..	**1**	**2**	**3**	**4**	**5**	**6**	**7**
6. (S)he is spending time with someone (s)he does *not* know that her / his own age? ……..	**1**	**2**	**3**	**4**	**5**	**6**	**7**

7. Someone familiar that is her / his own age walks up to her /him? …….	**1**	**2**	**3**	**4**	**5**	**6**	**7**
8. (S)he has just been separated from her / his main caregiver to be with an adult (s)he does *not* know? …….....	**1**	**2**	**3**	**4**	**5**	**6**	**7**
9. An adult (s)he does *not* know walks up to her / him? ……	**1**	**2**	**3**	**4**	**5**	**6**	**7**
10. (S)he is the focus of attention in a group of people her / his own age that (s)he does *not* know? ……...	**1**	**2**	**3**	**4**	**5**	**6**	**7**
11. (S)he is spending time with someone familiar that is her / his own age? ……	**1**	**2**	**3**	**4**	**5**	**6**	**7**
12. (S)he is the focus of attention in a group of people her / his own age that (s)he knows? …	**1**	**2**	**3**	**4**	**5**	**6**	**7**

How would the person you care for appear if…	**Very shy**	**Moderately shy**	**A little shy**	**Neither**	**A little sociable**	**Moderately sociable**	**Very sociable**
13. (S)he is with her / his main caregiver and then someone her / his own age that (s)he does *not* know starts to talk to her / him? …….....	**1**	**2**	**3**	**4**	**5**	**6**	**7**
14. A familiar adult walks up to her / him? …….....	**1**	**2**	**3**	**4**	**5**	**6**	**7**
15. (S)he is with her / his main caregiver and then an adult (s)he does *not* know starts to talk to her / him? ……..	**1**	**2**	**3**	**4**	**5**	**6**	**7**
16. (S)he is spending time with her / his main caregiver? ……..	**1**	**2**	**3**	**4**	**5**	**6**	**7**
17. (S)he is the focus of attention in a group of adults (s)he does *not* know? …….....	**1**	**2**	**3**	**4**	**5**	**6**	**7**

	Never or very rarely	**Rarely**	**Sometimes**	**About half the time**	**Often**	**Very often**	**Nearly Always**
18. When there are only familiar people around, how often does (s)he try to make contact with them in any way (by talking, signing, vocalising, using gestures, moving towards them in any way etc.)? …….	**1**	**2**	**3**	**4**	**5**	**6**	**7**
19. When familiar people and people are around who (s)he does *not* know, how often does (s)he try to make contact with the people (s)he does *not* know in any way (by talking, signing, vocalising, using gestures, moving towards them in any way etc.)? ……	**1**	**2**	**3**	**4**	**5**	**6**	**7**
20. When familiar people and people are around who (s)he does *not* know, how often does (s)he try to make contact with the familiar people in any way (by talking, signing, vocalising, using gestures, moving towards them in any way etc.)? ……	**1**	**2**	**3**	**4**	**5**	**6**	**7**

21. When there are only people around who (s)he does *not* know, how often does (s)he try to make contact with them in any way (by talking, signing, vocalising, using gestures, moving towards them in any way etc.)?

…….. **1 2 3 4 5 6 7**

22. Does the person you care for speak or sign **more** than 30 words? YES ☐ NO ☐

If you answered **'yes'** to this question, please complete the rest of the questionnaire. If you answered **'no'**, please complete the box at the end of the questionnaire if there is anything else you think we should know.

23. Does the person speak *less* than (s)he used to? ☐ ☐

24. Does the person *only* speak or sign in some settings and not others? ☐ ☐

If 'yes' please describe

.....

.....

.....

25. Does the person *only* speak or sign to some people and not others? ☐ ☐

If 'yes' please describe

……

……

……

Is there anything else you want to tell us about how the person you care for appears in social situations with other people (s)he knows or doesn't know, when separated from you, in a group setting or is the centre of attention in a group of people?

……...……...…..……

……..……...……

……...……..... .....

Thank you, please check your answers and go on to the next questionnaire.

Health Questionnaire

PART A

Instructions:

- Have these problems **EVER** affected your child or person you care for?
- Please rate as **0** – if the problem has never affected the person you care for, **1** – if it has been a mild problem, **2** - if the problem has been moderately serious, or **3** – if the problem has been severe.
- If the person you care for has had these problems please state whether any treatment has been implemented by circling yes

	Never	Mild	Mode rate	Severe
1a. Eye Problems (e.g. glaucoma / blocked tear duct/s).....	**0**	**1**	**2**	**3**
1b. Corrective surgery / medication / treatment: **yes / no**				
2a. Ear Problems (e.g. infections, glue ear)	**0**	**1**	**2**	**3**
2b. Corrective surgery / medication / treatment (e.g. grommets): **yes / no**				
3a. Dental Problems (e.g. toothache / gum problems / mouth ulcers / delayed eruption of teeth).....	**0**	**1**	**2**	**3**
3b.Dental surgery / treatment (e.g. teeth removal): **yes / no**				
4a. Cleft Palate.....	**0**	**1**	**2**	**3**
4b. Repaired: **yes / no**				
5a. Gastrointestinal Difficulties (e.g. reflux / stomach problems).....	**0**	**1**	**2**	**3**
5b. Corrective surgery / medication / treatment (e.g. nissen fundoplication): **yes / no**				
6a. Bowel Problems (e.g. obstruction).....	**0**	**1**	**2**	**3**
6b. Corrective surgery / treatment: **yes / no**				
7a. Heart Abnormalities or Circulatory Problems (e.g. congenital heart lesions or murmur).....	**0**	**1**	**2**	**3**
7b. Corrective surgery / medication / treatment: **yes / no**				
8a. Problems with Genitalia (e.g. prostate/ testicular problems i.e. undescended testes) ……..	**0**	**1**	**2**	**3**
8b. Corrective surgery / treatment: **yes / no**				
9a. Hernia (e.g. inguinal or hiatal).....	**0**	**1**	**2**	**3**
9b. Repair / treatment: **yes / no**				
10. Limb Abnormalities (e.g. malformed arm).....	**0**	**1**	**2**	**3**
11a. Epilepsy / Seizures / Neurological Referrals.....	**0**	**1**	**2**	**3**
11b. Medication: **yes / no**				
12a. Lung or Respiratory Problems (asthma/bronchitis).....	**0**	**1**	**2**	**3**
12b. Corrective surgery / medication / treatment: **yes / no**				
13a. Liver or Kidney Problems.....	**0**	**1**	**2**	**3**
13b. Corrective surgery / medication / treatment: **yes / no**				
14a. Diabetes or Thyroid Function Problems.....	**0**	**1**	**2**	**3**
14b. Corrective surgery / medication / treatment: **yes / no**				
15a. Skin Problems (e.g. tinea, eczema, psoriasis, dry skin)…….....	**0**	**1**	**2**	**3**
15b. Medication / treatment: **yes / no**				
16a. Other (please specify problem, severity from 0-3).....	**0**	**1**	**2**	**3**
16b. Corrective surgery / medication / treatment: **yes / no**				

PART B

Instructions:

- Have these medical problems affected the person you care for in the past **MONTH**

- Please rate as **0** – if your child has not been affected by this problem in the past month, **1** - if they have been mildly affected, **2** – if the problem has moderately affected your child and **3** - if your child has been severely affected by the problem.

	No	Mild	Moderate	Severe
17. Eye Problems (e.g. glaucoma / blocked tear duct/s).....	**0**	**1**	**2**	**3**
18. Ear Problems (e.g. infections, glue ear).....	**0**	**1**	**2**	**3**
19. Dental Problems (e.g. toothache / gum problems / mouth ulcers / delayed eruption of teeth).....	**0**	**1**	**2**	**3**
20. Cleft Palate.....	**0**	**1**	**2**	**3**
21. Gastrointestinal Difficulties (e.g. reflux / stomach problems).....	**0**	**1**	**2**	**3**
22. Bowel Problems (e.g. obstruction).....	**0**	**1**	**2**	**3**
23. Heart Abnormalities or Circulatory Problems (e.g. congenital heart lesions or murmur)…… ……...	**0**	**1**	**2**	**3**
24. Problems with Genitalia (e.g. prostate / testicular problems i.e. undescended testes)....	**0**	**1**	**2**	**3**
25. Hernia (e.g. inguinal or hiatal).....	**0**	**1**	**2**	**3**
26. Limb Abnormalities (e.g. malformed arm).....	**0**	**1**	**2**	**3**
27. Epilepsy / Seizures / Neurological Referrals.....	**0**	**1**	**2**	**3**
28. Lung or Respiratory Problems (asthma / bronchitis).....	**0**	**1**	**2**	**3**
29. Liver or Kidney Problems.....	**0**	**1**	**2**	**3**
30. Diabetes or Thyroid Function Problems.....	**0**	**1**	**2**	**3**
31. Skin Problems (e.g. tinea, eczema, psoriasis, dry skin).....	**0**	**1**	**2**	**3**
32. Other (please specify problem and severity from 0-3) ……..	**0**	**1**	**2**	**3**

Thank you, please check your answers and go on to the next questionnaire.

WESSEX Questionnaire

These items refer to the person you care for. For each question (A, B, C, D etc …), please enter the appropriate code in each box.

(Frequently = more than once a week)

A) Wetting (nights) **1** = frequently **2** = occasionally **3** = never ☐

B) Soiling (nights) **1** = frequently **2** = occasionally **3** = never ☐

C) Wetting (days) **1** = frequently **2** = occasionally **3** = never ☐

D) Soiling (days) **1** = frequently **2** = occasionally **3** = never ☐

E) Walk with help **1** = not at all **2** = not up stairs **3** = up stairs and elsewhere ☐

(note: if this person walks *by himself* upstairs and elsewhere, please also code '3' for 'walk with help')

F) Walk by himself **1** = not at all **2** = not up stairs **3** = up stairs and elsewhere ☐

G) Feed himself **1** = not at all **2** = with help **3** = without help ☐

H) Wash himself **1** = not at all **2** = with help **3** = without help ☐

I) Dress himself **1** = not at all **2** = with help **3** = without help ☐

J) Vision **1** = blind or almost **2** = poor **3** = normal ☐

K) Hearing **1** = deaf or almost **2** = poor **3** = normal ☐

L) Speech **1** = never a word **2** = odd words only **3** = sentences and normal **4** = can talk but doesn't ☐

If this person talks in sentences, is his/her speech:

1 = Difficult to understand even by acquaintances, impossible for strangers?

2 = Easily understood for acquaintances, difficult for strangers?

3 = Clear enough to be understood by anyone? ☐

M) Reads **1** = nothing **2** = a little **3** = newspapers and/or books

N) Writes **1** = nothing **2** = a little **3** = own correspondence ☐

O) Counts **1** = nothing **2** = a little **3** = understands money values ☐

Thank you, please check your answers and go on to the next questionnaire.

THE REPETITIVE BEHAVIOUR QUESTIONNAIRE (RBQ)

INSTRUCTIONS

1. The questionnaire asks about 19 different behaviours.
2. Each behaviour is accompanied by a brief definition and examples. The examples given for each behaviour are not necessarily a complete list but may help you to understand the definitions more fully.
3. Please read the definitions and examples carefully and circle the appropriate number on the scale to indicate how frequently the person you care for has engaged in each of the behaviours **within the last month**.
4. If a particular behaviour does not apply to the person you care for because they are not mobile or verbal please circle the number 0 on the scale

	Never	Once a month	Once a week	Once a day	More than once a day
1. Object stereotypy: repetitive, seemingly purposeless movement of objects in an unusual way *E.g. twirling or twiddling objects, twisting or shaking objects, banging or slapping objects.*	**0**	**1**	**2**	**3**	**4**
2. Body stereotypy: repetitive, seemingly purposeless movement of whole body or part of body (other than hands) in an unusual way. *E.g. body rocking, or swaying ,or spinning, bouncing, head shaking, body posturing.*. Does not include self-injurious behaviour.	**0**	**1**	**2**	**3**	**4**
3. Hand stereotypy: repetitive, seemingly purposeless movement of hands in an unusual way. *E.g. finger twiddling, hand flapping, wigging or flicking fingers, hand posturing.* Does not include self-injurious behaviour.	**0**	**1**	**2**	**3**	**4**
4. Cleaning: Excessive cleaning, washing or polishing of objects or parts of the body *E.g. polishes windows and surfaces excessively, washes hands and face excessively,*	**0**	**1**	**2**	**3**	**4**
5. Tidying up: Tidying away any objects that have been left out. This may occur in situations when it is inappropriate to put the objects away. Objects may be put away into inappropriate places. *E.g. putting cutlery left out for dinner in the bin, removes all objects from surfaces.*	**0**	**1**	**2**	**3**	**4**
6. Hoarding: Collecting, storing or hiding objects to excess, including rubbish, bits of paper, and pieces of string or any other unusual items.	**0**	**1**	**2**	**3**	**4**
7. Organising objects: Organising objects into categories according to various characteristics such as colour, size, or function. *E.g. ordering magazines according to size, ordering toy cars according to colour, ordering books according to topic.*	**0**	**1**	**2**	**3**	**4**
8. Attachment to particular people: Continually asking to see, speak or contact a particular 'favourite' person. *E.g. continually asks to see or speak to particular friend, carer, babysitter or schoolteacher.*	**0**	**1**	**2**	**3**	**4**
9. Repetitive questions: Asking specific questions over and over. *E.g. always asking people what their favourite colour is, asking who is taking them to school the next day over and over*	**0**	**1**	**2**	**3**	**4**

10. Attachment to objects: Strong preference for a particular object to be present at all times. *E.g. Carrying a particular piece of string everywhere, taking a particular red toy car everywhere, attachment to soft toy or particular blanket.*	**0**	**1**	**2**	**3**	**4**

	Never	**Once a month**	**Once a week**	**Once a day**	**More than once a day**
11. Repetitive phrases/signing: Repeating particular sounds, phrases or signs that are unrelated to the situation over and over. *E.g. repeatedly signing the word 'telephone'.*	**0**	**1**	**2**	**3**	**4**
12. Rituals: carrying out a sequence of unusual or bizarre actions before, during or after a task. The sequence will always be carried out when performing this task and will always occur in the same way. *E.g. turning round three times before sitting down, turning lights on and off twice before leaving a room, tapping door frame twice when passing through it.*	**0**	**1**	**2**	**3**	**4**
13. Restricted conversation: Repeatedly talks about specific, unusual topics in great detail. *E.g. conversation restricted to: trains, buses, dinosaurs, particular film, country, or sport.*	**0**	**1**	**2**	**3**	**4**
14. Echolalia: Repetition of speech that has either just been heard or has been heard more than a minute earlier. *E.g.: Mum:' Jack don't do that' Jack: 'Jack don't do that'.*	**0**	**1**	**2**	**3**	**4**
15. Preference for routine: Insist on having the same household, school or work schedule everyday. *E.g. likes to have the same activities on the same day at the same time each week, prefers to eat lunch at exactly the same time every day, wearing the same jumper everyday.*	**0**	**1**	**2**	**3**	**4**
16. Lining up or arranging objects*: Arrangement of objects into lines or patterns E.g. placing toy cars in a symmetrical pattern, precisely lining up story books,*	**0**	**1**	**2**	**3**	**4**
17. Just right behaviour: Strong insistence that objects, furniture and toys always remain in the same place. *E.g. all chairs, pictures and toys have a very specific place that cannot be changed.*	**0**	**1**	**2**	**3**	**4**
18. Completing behaviour: Insists on having objects or activities 'complete' or 'whole' *E.g. Must have doors open or closed not in between, story must be read from beginning to end, not left halfway through.*	**0**	**1**	**2**	**3**	**4**
19. Spotless behaviour: Removing small, almost unnoticeable pieces of lint, fluff, crumbs or dirt from surfaces, clothes and objects. *E.g. Picking fluff off a jumper, removing crumbs from the kitchen table.*	**0**	**1**	**2**	**3**	**4**

Thank you, please check your answers and go on to the next questionnaire.

Mood, Interest and Pleasure Questionnaire- Short-form (MIPQ-S)
Instructions for completing the MIPQ-S

This questionnaire contains 12 questions – you should complete all 12 questions. Each question will ask for your opinion about particular behaviours, which you have observed in the last 2 weeks. For every question you should circle the most appropriate response e.g.

6) In the last two weeks, how interested did the person appear to be in his/her surroundings?

interested all of the time	*interested most of the time*	*interested about half of the time*	*interested some of the time*	*never interested*

The Mood, Interest and Pleasure Questionnaire - Short Form

1) In the last two weeks, did the person seem…

Sad all of The time	sad most of the time	sad about half of the time	sad some of the time	never sad

Please comment if anything has happened in the last two weeks which you feel might explain sadness if it has been observed (e.g. a bereavement):

2) In the last two weeks, how often did you hear positive vocalizations* when the person was engaged in activities*?

all of the time	most of the the time	about half of the time	some of the time	never

*positive vocalizations: e.g. laughing, giggling, "excited sounds" etc.
*engaged in activities: i.e. when someone is actively involved in any activity such as a mealtime, a social interaction, a self-care task or social outing etc.

3) In the last two weeks, do you think the facial expression of the person looked "flat"*…

all of the time	most of the the time	about half of the time	some of the time	never

*flat expression: expression seems lifeless; lacks emotional expression; seems unresponsive.

4) In the last two weeks, would you say the person…

cried every day	cried nearly every day	cried 3-4 times each week	cried once or twice each week	cried less than once each week

5) In the last two weeks, how interested did the person appear to be in his/her surroundings?

interested all of the time	interested most of the time	interested about half of the time	interested some of the time	never interested

6) In the last two weeks, did the person seem to have been enjoying life…

all of the time	most of the the time	about half of the time	some of the time	never

Please comment if there are any reasons why this person might not have been enjoying him/herself e.g. illness, being in pain, experiencing a loss etc.:

7) In the last two weeks, would you say the person smiled…

at least once every day	at least once nearly every day	3-4 times each week	once or twice each week	less than once each week

8) In the last two weeks, how disinterested did the person seem to be in his/her surroundings?

disinterested all of the time	disinterested most of the time	disinterested about half of the time	disinterested some of the time	never disinterested

9) In the last two weeks, when the person was engaged in activities*, to what extent did his/her facial expressions* suggest that s/he was interested in the activity?

interested all of the time	interested most of the time	interested about half of the time	interested some of the time	never interested

*****engaged in activities: i.e. when someone is actively involved in any activity such as a mealtime, social interaction, self-care task or social outing etc.
*****facial expressions: interest might be indicated by the degree to which the person's gaze is being directed at the person/things involved in an activity.

10) In the last two weeks, would you say that the person…

laughed every day	laughed nearly every day	laughed 3-4 times each week	laughed once or twice each week	laughed less than once each week

11) In the last two weeks, how often did you see gestures which appeared to demonstrate enjoyment* when the person was engaged in activities*?

all of the time	most of the the time	about half of the time	some of the time	never

*gestures which appear to demonstrate enjoyment: e.g. clapping, waving hands in excitement etc.
*****engaged in activities: i.e. when someone is actively involved in any activity such as a meal time, social interaction, self-care task or social outing etc.

12) In the last two weeks, did the person's vocalizations* sound distressed…

all of the time	most of the the time	about half of the time	some of the time	never

*****vocalizations: any words, noises or utterances.

Please feel free to make any additional comments about the behaviour of the person over the last two weeks (continue overleaf if necessary):

Thank you, please check your answers and go on to the next questionnaire.

THE CHALLENGING BEHAVIOUR QUESTIONNAIRE (CBQ)

1) Has the person shown self-injurious behaviour in the last month? (e.g. head banging, head-punching or slapping, removing hair, self-scratching, body hitting, eye poking or pressing).

Yes ☐ No ☐

If the behaviour has not occurred, please go to question 6. If the behaviour occurred in the past month please answer questions 2 to 5:

2) Place a tick next to the item for any of the following list of behaviours which the person displays in a repetitive manner (repeats the same movement/ behaviour twice or more in succession):

Hits self with body part (e.g. slaps head or face)…… ☐
Hits self against surface or object (e.g. bangs head on floor or table)…… ☐
Hits self with object…… ☐
Bites self (e.g. bites hand on wrist or arm)…….. ☐
Pulls (e.g. pulls hair or skin)……..... ☐
Rubs or scratches self (e.g. rub marks on arm or leg)……. ☐
Inserts finger or objects (e.g. eye poking)……..... ☐
Other form of self-injury, please specify:…… ☐

3) In the last month, for how long did the **longest** episode or burst of his behaviour last? *(Please circle one number)*

1	2	3	4	5
Less than a minute	Less than 5 minutes	Less than 15 minutes	Less than an hour	More than an hour

4) In the last month as a result of this behaviour, has physical contact or prevention or restraint by others been necessary e.g. blocking, taking objects from an individual, temporary restraint of an arm? *(Please circle one number)*

0	1	2	3	4
Never	At least once a month	At least once a week	At least once a day	At least once an hour

5) Think about how often this behaviour occurred in the last month. If there was no change and you watched the person now, then would you definitely see the behaviour:

1	2	3	4	5
By this time Next month	By this time next week	By this time tomorrow	In the next hour	In the next 15 minutes

6) Has the person shown physical aggression in the last month? (e.g. punching, pushing, kicking, pulling hair, grabbing other's clothing).

Yes ☐ No ☐

7) Has the person shown disruption and destruction of property or the environment in the last month? (e.g. tearing or chewing own clothing, tearing newspapers, breaking windows or furniture, slamming doors, spoiling a meal).

Yes ☐ No ☐

8) Has the person shown stereotyped behaviours in the last month? (e.g. rocking twiddling objects, patting or tapping part of the body, constant hand movements, eye pressing).

Yes ☐ No ☐

Thank you, please check your answers and go on to the next questionnaire.

FOOD-RELATED PROBLEMS QUESTIONNAIRE

These items refer to the person in your care and food-related issues. Please circle one number
only. If the person does not speak, please tick the box 'Does not apply.'

Please answer the following:

1. How often does the person compare the size or content of their meal with others?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

2. If given the opportunity, how often would the person 'help themselves' to food which they should not have?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

3. Is the person ever able to accept an explanation given to them if a meal is delayed?

6	5	4	3	2	1	0
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

4. Does the person ever hide or hoard food?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

5. How often does the person talk about food (Not applicable □)?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

6. If the person was denied food, how often would they respond negatively?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

7. Is it necessary to lock food away to stop the person from taking food?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

8. After a normal size meal, how often will the person say they still feel hungry (Not applicable □)?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

9. If the person was tired, ill or upset, how often would this result in them going without food?

6	5	4	3	2	1	0
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

10. If it was available, would the person eat food not suitable for consumption (eg. frozen food,
scraps from dustbins)?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

11. If a meal includes an item of food the person does not like or is not expecting, how often would this result in behavioural difficulties?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

12. Does the person ever eat non-edible items (e.g. dog food, leaves)?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

13. How frequently will the person share food with others?

6	5	4	3	2	1	0
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

14. Does the person ever describe 'feeling full' (Not applicable □)?

6	5	4	3	2	1	0
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

15. Does the person ever associate people and/or places with specific food items or occasions involving food?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

16. If given the opportunity, does the person ever eat more than a standard sized meal?

0	1	2	3	4	5	6
Never	Almost never	Seldom	Half the time	Usually	Almost always	Always

17. Please describe the extent to which (if at all) external control is needed to be in place in an effort to stop the person taking food. If controls are in place, please describe (e.g. locked kitchen, constant supervision):

18. Please describe the extent to which (if at all) the person demonstrates self-control around food and the ways in which they may exert control (e.g. asks for food items to be removed):

19. Any other comments about food-related issues:

Thank you very much for taking the time to complete these questionnaires.

Appendix 1b

List of toys employed in the social presses

Instructions for running the social presses

Behaviour descriptions and coding schedule

Social press toy list

Younger/less able individuals:

Plastic blocks
Sponge balls
Animal puzzle
Animal blocks
Xylophone
Rain maker…(sorry…very noisy!)
Pop up key board
Doll from ADOS
Big Truck from ADOS
Peek-a-boo blanket from ADOS

Older/more able individuals:

Hard back book
Magnetic alphabet board
Magic writer
Slinky
Jitter ball – in place of soft ball throw and catch
Wooden farm set
Animal snap cards – in place of tower building
Binoculars
Pens
Lace board
Pin- art
Small game

Bubble gun

Protocol for structured social presses

The structured social presses will take place in a quiet and safe environment, such as an empty classroom. Conditions 1, 2, 3 and 5 will be conducted for three minutes. Condition 4 will be conducted until all of the social presses have been completed (approx ten minutes). Present in each session will be one adult examiner and one/two people operating a video recorder and/or prompting the examiner through the presses. The three conditions will be administered once by an adult stranger and once by a familiar adult (person who works regularly with the participant and has known them for at least 3 months, parent, carer or teaching staff). There are two versions of the structured social presses assessment. The first is for younger or less able individuals (SS1). The second (SS 2) version has been modified for older or more able individuals for whom the toys and tasks in SS 1 are not appropriate. Unless stated – the instructions for the conditions should be followed for both SS1 and SS2 individuals.

The sessions will be video recorded for later analysis. The privacy and dignity of the participants will be respected at all times.

Condition 1: 'Warm up' condition: 'Hello….look at all of these toys. Let's play together' – two minutes

1. Free play toys/activities as listed in Appendix 1b are present. All toys/activities are visible and accessible to the participant.

2. Adult plays/engages with the participant for **two minutes** in any way. The adult should attempt to obtain the participant's eye contact or attention in some way before starting condition 1 in order to ensure that the participant is aware of the adult's presence and availability for interaction.

3. Adult provides verbal and physical attention to the child throughout.

Condition 2: Response Engagement: 'I'm going to sit here with you, but you play with the toys on your own for a while'- three minutes

1. Free play toys/activities as listed in Appendix 1b are present. All toys/activities are visible and accessible to the participant.

2. The adult is seated within close proximity of the participant (i.e. visible and within touching distance of the participant). The adult looks and smiles at the participant but does not initiate any interaction with the participant or direct the participant's behaviour in any way.

3. If the participant approaches the adult and initiates interaction, the adult responds appropriately by describing the participant's behaviour or following any requests made by the participant. After five seconds the adult terminates this engagement. Return to point 2.

Condition 3: Non-engagement: 'I'm going to talk to …..now for a while, you play with the toys' – three minutes

1. Free play toys/activities as listed in Appendix 1b are present. All toys/activities are visible and accessible to the participant.

2. The adult is seated within close proximity of the participant (i.e. visible and within touching distance of the participant). The adult does not look, smile or talk to the participant. The adult does not initiate interaction with the participant and does not respond to the participant's attempts to engage with them. **NB: For older/more able individuals it may be more comfortable for the adult to indicate that they are making some notes/reading a book rather than talking to the other researcher.**

3. If the participant moves away from the adult, the adult should remain seated.

As far as possible the participant should be ignored with no consequences for aggressive behaviour. Where intervention is necessary this should be brief and done with limited eye contact and verbal response.

Condition 4: Active Engagement: 'Can I join in with you now?' – continue until hierarchy of demands has been completed.

1. Free play toys/activities as listed in Appendix 1b are present. All toys/activities are visible and accessible to the participant.

2. The adult is seated within close proximity of the participant (i.e. visible and within touching distance of the participant). The adult looks, smiles and talks to the participant at least every three seconds. The adult should remain within close proximity of the participant at all times.

3. The adult actively engages with the participant and initiates interaction with the participant, describing the participant's behaviour and the objects they are playing with. The adult responds to all initiations made by the participant.

4.

   a. <u>For younger/less able individuals</u>, after 2 minutes of free play, the adult initiates a turn taking game using the plastic ball, by rolling it to and fro towards the participant. If the participant responds to this activity, the adult continues this engagement for two further sequences and then proceeds to point 5 (even if the participant only responds to the first initiation of the activity). If the participant does not respond to this activity after three attempts then proceed to point 5.

   b. <u>For older/more able individuals,</u> after 2 minutes of free play, the adult initiates a turn taking game using the 'jitter ball'. If the participant responds to this activity, the adult continues this engagement for two further sequences and then proceeds to point 5 (even if the participant only responds to the first initiation of the

activity). If the participant does not respond to this activity after three attempts then proceed to point 5.

5.

a. With younger/less able individuals, the adult initiates building a tower with blocks and asks the participant to help them build it as high as possible. When the tower is built the adult should pretend to knock the tower down. After two further repetitions of the activity proceed to point 6.

b. With older/ more able individuals, the adult initiates a game of snap with the animal snap cards. The game should continue for at least three matching cards. Following this or completion of the game, proceed to point 6.

6.

a. With younger/less able individuals, the adult initiates a social play activity such as a tickling game or peek-a-boo game with the participant. After two further repetitions of the activity proceed to point 7.

b. With older/more able individuals, the adult should informally initiate a relatively high demand social conversation with the participant, asking them about their hobbies, what they like to do at school, whether they have any brothers or sisters. During the conversation, the adult should offer some new information about themselves i.e. I went shopping yesterday ….they should pause briefly after this statement to allow the participant to comment. During the conversation the adult should also offer an interesting piece of information about themselves that the participant would be expected to comment on i.e. I've come from a very long way to see you today. The adult should be careful not to dominate the conversation and should allow plenty of time for the participant to respond or elaborate by asking both open ended questions (tell me about what you do at school?) and closed ended questions (do you like music lessons?). Allow at least one minute for conversation. After at least one minute of conversation, proceed to point 7.

7.

a. With younger/less able individuals, the adult introduces a new, toy the bubble gun, to the participant. The adult holds the pot of liquid and blows the bubbles so that they are clearly visible to the participant. The adult comments on the bubbles and encourages the participant to look at them, touch them or pop them. The adult then pauses, looks and smiles at the participant for five seconds. If the participant makes a request for the bubbles in some way by gesturing, vocalising, pointing or reaching, then the adult repeats this engagement in the same manner for two further sequences and then proceeds to point 8. If the participant wishes to hold the bubble gun themselves, the adult should keep hold of the pot of liquid and encourage the participant to seek the pot of liquid in order to activate the bubble gun. If the participant does not respond after three attempts to initiate interaction and does not appear to be motivated by this activity, select two play objects (if a preferred toy is observed during free play then use this as one of the options) and offer them a choice of either of these and wait for them to indicate which of these toys they would like. If the participant does not select a toy, offer

them the preferred toy to play with. Repeat the sequence with the same objects after 1 minute of play with the preferred object.

b. For older/more able individuals, Examiner brings out remote controlled car/toy and demonstrates its use. Adult comments on the object and encourages the participant to look at it. The adult then pauses, looks and smiles at the participant for five seconds. If the participant makes a request for the remote control or to see the object again, this sequence should be repeated. If not the remote should be placed in front of the participant thus allowing them to use the object as they wish.

8. The adult indicates that the session is going to end and asks the participant to help them to put the toys/activities away. The adult begins to put the toys away into the box/bag in front of the participant.

Condition 5: Non-engagement no toys: 'I'm going to talk to … again for a few minutes.' – three minutes

1. The adult is seated within close proximity of the participant (i.e. visible and within touching distance of the participant). The adult turns away from the participant and talks to the researcher for the three minute period.

2. No toys/activities are present, the participant has no activities to occupy or distract them.

3. The adult does not look, smile or talk to the participant. The adult does not initiate interaction with the participant and does not respond to the participant's attempts to engage with them. **NB: For older/more able individuals it may be more comfortable for the adult to indicate that they are making some notes/reading a book rather than talking to the other researcher.**

4. If the participant moves away from the adult, the adult should remain seated.

Behaviour descriptions and coding schedule

	0	1	2	3	4
Positive emotional affect (Eg. positive facial expressions, vocalisations and manner such as smiling, laughing, clapping hands.)	No examples of positive affect at any stage.	Some examples of positive affect but only tentative or occasional.	Affect positive about half of the time. May consist of brief expressions of positive affect in response to particular activities for example, but affect not sustained between these instances.	Affect positive most of the time. May consist of brief expressions of positive affect in response to particular activities for example, but also sometimes sustained between these instances.	Affect generally positive throughout and often sustained between expressions of positive affect in response to particular activities.
Negative emotional affect (Eg. negative facial expressions, vocalisations and manner such as crying and frowning. Participant may appear distressed or angry.)	No examples of negative affect at any stage.	Some examples of negative affect but only tentative or occasional.	Affect negative about half of the time. May cry in response to particular activities for example, but affect not sustained between these instances.	Affect negative most of the time. May cry in response to particular activities for example, but also sometimes sustained between these instances.	Affect generally negative throughout and often sustained between expressions of negative affect in response to particular activities.
Frequency of spontaneous physical contact (Include all participant initiated physical contact, regardless of nature of contact or intent.)	No spontaneous physical contact initiated with the examiner or other adult.	One or two examples of spontaneous initiation of physical contact.	Several examples of spontaneous initiation of physical contact.	Spontaneous physical contact initiated frequently but not to an excessive or socially inappropriate degree.	Spontaneous physical contact initiated frequently to an excessive or socially inappropriate degree.
Nature of physical contact initiated (Rate nature of instances of spontaneous contact observed in previous item regardless of frequency.)	**EITHER** No spontaneous physical contact initiated with the examiner or other adult **OR** Contact mostly negative or aggressive in nature (e.g. hair pulling or hitting).	Physical contact mostly negative or aggressive in nature (e.g. hair pulling or hitting) but one or two instances of positive physical contact (e.g. hugging, climb onto lap, tapping to gain attention) also observed.	Contact generally neither negative nor positive in nature and does not appear socially motivated. May be for personal demands only such as to gain attention/ assistance or for sensory stimulation/interest (e.g. sniffing or peering at examiner)	Contact is mostly positive in nature (e.g. hug, climb onto lap, tapping to gain attention) but one or two instances of negative physical contact (e.g. hair pulling or hitting) also observed	Physical contact always positive in nature and appears socially motivated and affectionate (e.g. hug, climb onto lap, tapping to gain attention).
Social responsiveness * Responds to specific behavioural requests, suggestions, questions or their name (if used). ****** Elaboration is defined as when the participant spontaneously builds on what is expected of them e.g. the participant independently initiates building the block tower again once it has been knocked down.	Unresponsive and disinterested. Does not respond*. Largely ignores what the examiner is doing.	Unresponsive but some interest. May not respond* but attends to what examiner is doing (this must be more than a fleeting glance).	Interested and occasionally responsive. Responds* at least once but interactions are examiner led and are not progressive or reciprocal. Participant mostly attentive to examiner.	Interested and highly responsive. Responds* more often than not. Interactions progressive and reciprocal. At least one or two examples of a back and forth exchange of several steps but participant does not elaborate** beyond initial examiner suggestions.	Interested and elaborately responsive. Responds* more often than not. More than two examples of back and forth exchanges of several steps. Participant elaborates** on initial examiner suggestions.
Avoidance of social interaction ***** Aversion includes aversion to gaze or touch, turning back on examiner, pushing examiner's hand away or removing self from proximity of	Consistently shows aversion* to all examiner approaches.	Shows aversion* to most but not all examiner approaches.	Shows aversion* to about half the examiner's approaches.	Occasionally shows aversion* to examiner approaches.	Shows aversion* to none of the examiner approaches.

examiner.					
Social anxiety *Anxiety shown by e.g. rigid or tense posture or fixed expression, close monitoring of examiner behaviour.	Marked anxiety* throughout assessment.	Participant shows mild signs of anxiety* or self-consciousness throughout assessment.	Participant shows mild signs of anxiety* or self-consciousness at some points during assessment but not throughout.	Participant seems hesitant about some interactions but not overtly anxious*.	Participant does not appear anxious* or hesitant at any stage.
Spontaneous initiation of interaction * Initiation of interaction may be verbal or non-verbal (e.g. approaching the examiner, offering or requesting objects, speaking or signing, touching the examiner to attempt to gain their attention (aggressively or otherwise), gesturing or pointing to an object while looking at examiner.	No clear spontaneous initiation of interaction* with examiner.	One or two examples of spontaneous initiation of interaction* with examiner but for personal demands or other unclear purpose only.	Three or more examples of spontaneous initiation of interaction* with the examiner but for personal demands or unclear purpose only	One or two examples of spontaneous initiation of interaction*with the examiner which appears to be socially motivated (e.g. for the purpose of being friendly) and not merely for personal demands (e.g. giving or showing an object).	More than three examples of spontaneous initiation of interaction* with the examiner which appear to be socially motivated (e.g. for the purpose of being friendly) and not merely for personal demands (e.g. giving or showing an object).
Focus of attention (objects focus vs. people focus)	Focus of the participant's attention either unclear or entirely object focussed. Participant does not attend to or show any interest in other people.	Focus of the participant's attention mostly on objects. Some attention paid to other people even if only for monitoring purposes.	Focus of the participant's attention shared between people and objects.	Focus of the participant's attention mostly on people. Attention appears to be socially motivated at least some of the time and not simply for purpose of monitoring.	Focus of the participant's attention almost entirely on people perhaps to an excessive degree. Attention appears to be mostly socially motivated.
Motivation for adult engagement * Interaction may be verbal or non-verbal (e.g. approaching the examiner, offering or requesting objects, speaking or signing, touching the examiner to attempt to gain their attention (aggressively or otherwise), gesturing or pointing to an object while looking at examiner.	The participant does not attempt to initiate interaction*. Either sits passively or entertains self (e.g. plays with the toys or passively listens to conversation between examiners).	The participant makes one or two attempts to initiate interaction* but when attention is not given they give up quickly and entertain self.	The participant makes three or more attempts to initiate interaction* but eventually gives up and entertains self. May subsequently return for renewed attempt to engage with adult but there must be a clear gap in their efforts.	The participant makes persistent attempts to initiate interaction* throughout the observation but stays within socially appropriate limits (e.g. approaching, vocalising (not high volume), touching hand or tapping shoulder).	The participant makes persistent attempts to initiate interaction* throughout the observation and through several different means, to the extent of using socially inappropriate methods (e.g. face grabbing, physical aggression such as hair pulling or through engaging in challenging behaviour).
Frequency of eye contact *Eye contact defined as the participant looking up/at the examiner, fixating on their eyes or face.	No eye contact* made with examiner.	Occasional, fleeting eye contact* made with examiner.	Frequent, fleeting eye contact* made with examiner	Frequent fleeting **AND** occasionally sustained eye contact* made with examiner.	Frequent, sustained eye contact* made with examiner.
Nature of eye contact * Appropriate integration of eye contact with other social-communication skills including gesture, pointing or facial expressions e.g. participant checks what examiner is looking at, or points, then follows examiner's gaze to check point has been registered. *Inappropriate eg. staring or avoidant.	No eye contact made with examiner.	Eye contact obviously awkward or inappropriate* in nature on all occasions - not naturally integrated with other behaviours during interaction. Includes participants who show prolonged eye contact (e.g. staring)	Eye contact somewhat awkward or inappropriate* in nature - not naturally integrated with other behaviours on every occasion but on some.	Eye contact slightly awkward or inappropriate* in nature - mostly naturally integrated with other behaviours during interaction but not always.	Eye contact consistently naturally and appropriately integrated* with other behaviours during social interaction.
Social communication style (Rate the majority of examples of social-	Little or no verbal or non-verbal communication at all.	Some vocalisations or gestures mostly indicating	Some clearly communicative vocalisations (verbal or non-	Some clearly communicative vocalisations (verbal or non-	Regular clear speech and or signing (e.g. BSL or

communication demonstrated by the participant rather than the best example)		affect (e.g. laughing or crying sounds indicating excitement) and not specifically communicative or directed at others **AND/OR** attempts to communicate through grabbing/touching or other physical means that has clear communicative intent.	verbal) or gestures (e.g. pointing, nodding and shaking head). Makes attempts to communicate specific desires but does not use speech or signing.	verbal) or gestures (e.g. pointing, nodding and shaking head). Makes attempts to communicate specific desires **AND** shows some use speech or signing which may be infrequent or unclear.	Makaton). Makes attempts to communicate specific desires which may be for the purpose of being friendly or otherwise.
Quality of social communication skills * Communication of simple desires may include indicating desire for attention, for assistance such as to be lifted up or desire for an object.	The participant either rarely attempts to communicate or makes attempts to communicate which cannot be understood.	The participant's attempts to communicate are often difficult to understand but they are occasionally able to communicate simple* desires.	The participant's attempts to communicate are sometimes difficult to understand but they are mostly able to communicate simple* desires and sometimes more complex desires, ideas and thoughts.	The participant is mostly able to communicate even complex desires, ideas and thoughts although to someone who does not know them well it is not always easy to understand them (e.g. they may have problems with articulation).	The participant is able to communicate and it is easy to understand their intentions and desires.

Appendix 2

Public Domain Briefing Document

An investigation of sociability: Delineating a behavioural and social phenotype for Monosomy 1p36 Deletion Syndrome

The research was conducted by Fay Cook from the School of Psychology at the University of Birmingham as part of the Doctorate in Clinical Psychology (Clin. Psy. D) training programme.

Outline

Literature Review

Research has shown that compromised social functioning for individuals with intellectual disabilities can have far reaching implications for quality of life, community participation and well being. The body of research on this topic is vast, but is affected by the lack of definitions for key social concepts. A literature review explored the commonly used definitions of four social concepts: social cognition, social competence, social skills and social behaviour; in both the wider cognitive and social psychology literature and in research on individuals with intellectual disabilities. It was found that few definitions exist and none are universally accepted and applied in research. Potential working definitions for three of the concepts were suggested. The review calls for further research in the area but also for researchers to define the social concepts they are investigating so definitional problems can be eradicated.

Empirical Paper

Background

Monosomy 1p36 Deletion Syndrome is a rare genetic disorder which results in a number of clinically identifiable characteristics, including facial, skeletal, neurological and developmental features. However, whilst the physical characteristics of the condition are well documented,

behavioural aspects are not, although some features may form a behavioural phenotype of 1p36. A behavioural phenotype has been defined as "the heightened probability or likelihood that people with a given syndrome will exhibit certain behavioural and developmental sequelae relative to those without the syndrome"(Dykens, 1995). The features which appear pertinent to 1p36 are aggression, temper tantrums, throwing or banging objects, striking people, screaming, self-injury and autistic features.

Social aspects of the condition have also been neglected although there are indications that individuals with 1p36 syndrome have certain difficulties, including reduced social interaction, repetitive stereotypies (e.g. holding hands in front of face, hand washing or flapping), tendency to beat, smell or roll objects repetitively, manual apraxia (twisting hands in a washing manoeuvre) and poor eye contact. However, as social aspects have not been the focus of research efforts, no consistent assessments or methods of obtaining information on social aspects of the syndrome have been applied. Therefore standardised assessments of social and behavioural aspects of the condition would allow the process of describing the behavioural and social aspects of 1p36.

Once standardised assessment methods are applied to 1p36 syndrome it will be possible to understand the social aspects of the condition with reference to other genetic syndromes with well defined phenotypes e.g. Angelman syndrome, Oliver, Berg, Moss, Arron & Burbidge, in review; Cornelia de Lange syndrome, Moss, Kaur, Jephcott, Berg, Cornish & Oliver, 2008; Oliver, Arron, Hall & Sloneem,2008; Richards, Moss, O'Farrell & Oliver, in press). By comparing Monosomy 1p36 deletion syndrome to other syndromes known for excessive sociability or social deficits where a behavioural and social phenotype has been described, it will be possible to describe and understand the behavioural and social phenotypes of 1p36.

There is a difficulty, however, with only employing standardised assessment measures, as these may not capture the nuances of sociability or social interactions. Therefore, methods which can measure and categorise the social traits and which can systematically alter the conditions under

which social traits may be expressed, may provide a rich source of information about sociability in 1p36 syndrome. Such a method combined with standardised assessments may provide a good starting point from which to begin to characterise the social phenotype of the syndrome.

Aims

The primary aim of the current research is to begin the process of characterising the behavioural phenotype with a focus on the social phenotype for Monosomy 1p36 deletion syndrome and compare this to other genetic syndromes (Angelman syndrome, Cornelia de Lange syndrome and Cri du Chat syndrome). By employing measures that are appropriate, reliable and valid for individuals with intellectual disabilities (standardised assessments) and incorporating a method of experimentally altering the social conditions under which children are observed interacting with others (social press), the research will extend existing literature.

Participants

90 participants included in the comparison study, 23 with 1p36 syndrome, Cornelia de Lange and Cri du Chat syndrome and 21 with Angelman syndrome; with an age range of eighteen months to forty five-years-old. In the social press study twelve individuals with 1p36 were included, age range three years thirteen months to thirteen years eleven months.

Design, Procedure and Coding

Comparison study: Parents and carers of individuals with 1p36 completed a number of standardised questionnaires on aspects of behaviour and sociability. The information from these was then compared to that held on the three other genetic syndromes.

Social press: Individuals were observed interacting with a familiar and unfamiliar adult where adult engagement/attention was manipulated across five conditions (two high engagement, 2 no engagement/response from the adult and one where the adult only responded to initiations of

interaction from the individuals). Video recordings of the observations were coded for social behaviours and skills in each of the five conditions, with familiar and unfamiliar adults.

Findings

Results from the comparative study indicate impaired social communication, lowered mood and higher sociability with familiar adults are all notable characteristics for 1p36. In the social presses individuals were more social under conditions of high attention/engagement with both familiar and unfamiliar people. In the social presses no differences in sociability were found between interactions with familiar and unfamiliar adults.

Clinical Implications and Future Directions

The higher levels of sociability in individuals with 1p36 syndrome found under conditions of high engagement could be used by clinicians to improve the effectiveness of interventions for individuals with 1p36 syndrome. Clinicians should also be aware of the potential for individuals to be sociable even with unfamiliar adults, and perhaps build such knowledge into interventions such as social skills training. It is therefore important for clinicians to be aware of the behavioural and social aspects of 1p36 syndrome and not just the genetic and molecular features when working with families affected by the condition.

Future research should concentrate on investigating further the social aspects of the condition, particularly behaviours that could be classed as autistic features

Conclusions

The study is the first to investigate social behaviour in 1p36 syndrome and as such the conclusions drawn are tentative. There is evidence that some characteristics may form part of a behavioural and social phenotype for the condition.

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