VOLUME I

RESEARCH COMPONENT:
SELF INJURY IN 1P36 DELETION SYNDROME

BY

ABBY MARR

A thesis submitted to
The University of Birmingham
For the degree of
DOCTORATE OF CLINICAL PSYCHOLOGY

Department of Clinical Psychology
School of Psychology
The University of Birmingham
June 2009

Overview

This thesis is submitted in partial fulfilment of the requirements for the degree of Doctorate of Clinical Psychology (ClinPsyD) at the University of Birmingham. The thesis comprises a research component (Volume I) and a clinical component (Volume II)

Volume I consists of two papers, a literature review and an empirical research study. The literature review examines studies that investigated the assessment of hand mouthing in individuals with severe to profound intellectual or multiple disabilities. The aetiology and subsequent treatment of hand mouthing as a behaviour maintained by automatic reinforcement is discussed, with a focus on the interaction between biological and environmental determinants of hand mouthing. The empirical research study examined the prevalence and correlates of self-injurious and aggressive behaviour in children with 1p36 deletion syndrome. Results from a survey indicate that self-injury is common in 1p36 deletion syndrome and is associated with overactivity and stereotyped behaviour. Aggression was found to be associated with impulsivity and repetitive behaviour. Experimental functional analyses confirmed high levels of hand mouthing with evidence of attention maintained function of self-injury. The implications of this finding for treatment are discussed, with particular reference made to shaping precursor behaviours to have a communicative function prior to the development of self-injurious behaviour. These papers are prepared for submission to *Journal of Intellectual Disability Research.* Contrary to journal requirements, tables and figures have been integrated into the text.

Volume II contains five clinical practice reports presenting psychological work conducted during placements in the specialties of child, learning disabilities, adult and older people. The first report describes cognitive and systemic formulations of a ten year old female and her family who presented with anxiety difficulties. The second report contains a service evaluation exploring the views of looked-after young people on a psychology service. The third report is a single-case experimental design used to evaluate a behavioural intervention for toileting difficulties in an 8-year-old boy with Autism. The fourth report describes a cognitive-behavioural formulation and intervention for a 32-year-old woman with depression and anxiety. The final report is an oral presentation that describes a cognitive-behavioural formulation and life story work with a 93-year-old male suffering from low mood.

Acknowledgements

I would like to express my thanks to the following people:

Unique, The Rare Chromosome Disorder Support Group, for supporting this research, to the families and children who participated in the study. Without the interest and enthusiasm from these families this research would not have been possible.

To Fay, my research partner, for all of her support throughout this project and for making the research trips so much fun!

To Professor Chris Oliver for his support, ever evolving ideas and interest in the work and to Dr Jo Moss for her patience and statistical advice. To Penny Tunnicliffe for her swift supportive replies to emails and for all of her help with the project from start to finish.

Special thanks to my family and friends for all their support and encouragement throughout my many years of study. Finally, the work is finished.

Finally, to Malc, for his continuous interest, reassurance, humour and love, throughout.

Contents of Volume I

Appendices

List of Tables

List of Figures

Literature Review

Assessment and aetiology of hand mouthing in individuals with severe to profound intellectual and disability: A review of the literature.

Abstract

Hand mouthing can be a form of self-injurious behaviour and has been shown to have a considerable impact on the adaptive and social functioning of people with intellectual and developmental disabilities. This paper reviews studies that investigated the assessment of hand mouthing in individuals with severe to profound intellectual disability or multiple disability. A literature search identified 30 studies published between 1995 and 2009 that used experimental functional analysis to determine the reinforcing properties of hand mouthing in a total of 53 individuals. In the vast majority of individuals (47) hand mouthing was found to be automatically reinforced, that is, hand mouthing was not found to be maintained by environmental influences, such as social attention or escape from a demanding task. A brief review of the literature on treatments of hand mouthing is presented, with emphasis given to the implications of pre-intervention assessment. The aetiology of hand mouthing as a behaviour maintained by automatic reinforcement is discussed, with a focus on the interaction between biological and environmental determinants of hand mouthing. Given the findings from this literature review, direction for future work is suggested.

Introduction

Self-Injury

Self-injurious behaviour (SIB) is defined as:

> 'Any behaviour, initiated by the individual, which directly results in physical harm to that individual.' (Murphy & Wilson, 1985; pg.15)

SIB includes, for example, head punching, hitting or banging against objects, self-biting, hitting other body parts and skin picking (Cooper, Smiley, Allan, Jackson, Finlayson, Mantry & Morrison, 2009). SIB behaviour is shown by around 4-12% of children with severe intellectual disability. There is considerable variability in the reported prevalence among adults with intellectual disabilty, with rates ranging from as low as 1.7% (Rojahn, 1986) to as high as 41% (Saloviita, 2000). A more recent study of 1023 adults with intellectual disabiity estimated prevalence to be 4.9% (Cooper et al., 2009). Prevalence and severity of SIB increase with age and peak during the early to mid 20's (Oliver, Murphy & Corbett, 1987). The limited research into onset indicates that SIB may begin as early as under 5 years of age (Berkson, Tupa and Sherman, 2001) and data on chronicity show that SIB can become a longstanding problem (Kebbon and Windahl, 1986; Emerson, Kiernan, Alborz, Reeves, Mason & Swarbrick et al., 2001). SIB can have a significant impact on the quality of life of the person and their family and can result in a need for long-term support from specialist services (Cooper et al., 2009; Emerson et al., 2001). One particular behaviour which can be considered a form of SIB, that has received attention in the research literature, is hand mouthing.

Hand mouthing

Hand mouthing has been generally defined as any response that results in getting the hands and face wet due to contact with saliva and the prevalence has been estimated to be between 7 and 16% for individuals with severe to profound developmental disabilities. (Rast & Jack, 1992). The behaviour has also been defined by topography, for example: *hand mouthing,* defined as inserting the hand past the plane of the lips and not closing the teeth on the hand; *hand biting*, defined as inserting the hand past the plane of the lips and closing the teeth on the hand and *finger sucking,* defined as inserting the finger past the plane of the lips and not closing the teeth on the finger (Cannella, O'Reilly & Lancioni, 2006).

Research findings have shown that engaging in hand-mouthing can have a number of detrimental effects on an individual's health, adaptive functioning and social interactions. (Pelios, Morren, Tesch & Axelrod, 1999; Wallace, Iwata, Zhou & Goff, 1999; Canella et al., 2006). Continuous hand-mouthing can lead to a number of physical health difficulties including tissue damage, increased risk of infection, bruising and scarring (Ball, Campbell & Barkmeyer, 1980; Fischer, Iwata & Mazaleski, 1997; Realon, Favell & Cacace, 1995). Hand-mouthing has been found to interfere with an individual's ability to participate in learning opportunities, due to their hands being consistently in the mouth, meaning they are unable to explore their environment and participate in educational and daily activities (McClure, Moss, McPeters & Kirkpatrick, 1986). Researchers have also found that hand-mouthing can limit social interactions and can become a barrier to social integration due to the production of offensive odours and sights. (Fischer et al., 1997; Lancioni, Singh, O'Reilly, Sigafoos, Oliva, Pidala et al., 2007; McClure et al., 1986; Realon et al., 1995). Investigation of hand mouthing has focused not only on the consequences for the individual, but also on the assessment, cause and intervention (e.g. Canella et al., 2005; Goh, Iwata, Shore, DeLeon, Lerman, Ulrich & Smith, 1995). One strategy for exploring the aetiology of hand mouthing has been to examine hand mouthing in genetic syndromes in which the prevalence of the behaviour is suggested to be elevated.

Hand mouthing and genetic syndromes

A number of studies have highlighted that individuals with particular genetic sydromes are at risk for developing self-injurious behaviour and in particular, hand-mouthing, for example, Fragile X, Rett, Lesch-Nyhan and Cornelia de Lange syndromes (Arron, Oliver, Berg, Moss & Burbidge, in press; Berney, Ireland, & Burn, 1999; Christie, Bay, Kaufman, Bakay, Borden & Nyhan, 1982; Largo & Schinzel, 1985; Hall, Oliver & Murphy, 2001; Lesch & Nyhan, 1964; Moss, Oliver, Hall, Arron, Sloneem & Petty, 2005; Oliver, Murphy, Crayton & Corbett, 1986; Steinhausen, von Gontard, Spohr, Hauffa, Eiholzer & Backes et al., 2002; Symons, Clark, Hatton, Skinner & Bailey, 2003)

Researchers have reported SIB to occur in between 38% to 58% of individuals with Fragile X Syndrome (FXS), with hand biting the most frequent topography (Largo & Schinzel, 1985; Steinhausen et al., 2002; Symons et al., 2003). Research into SIB in FXS has indicated that environmental factors may contribute to the development and maintenance of SIB. For example, in their survey of 55 young boys with FXS, Symons at al. (2003), found SIB to be

reported in 58% of the individuals and was most likely to occur following the presentation of difficult task demands or changes in routine.

In individuals with Rett syndrome (RS), the most prominent behavioural feature involves stereotypic, repetitive hand movements that are often categorised as self-injurious, including self-scratching, mouth hitting, hand biting and hand mouthing (Iwata, Pace, Willis, Gamache & Hyman, 1986; Oliver et al., 1986; Sansom, Krishnan, Corbett & Kerr, 1993). Research has demonstrated that up to 49% of individuals with RS exhibit at least one topography of SIB with the majority of behaviours involving the hands (Sansom et al., 1993). Roane et al. (2001) suggest that the high prevalence of these behaviours in individuals with RS indicates that the development of these behaviours may be related to organic causes. However, research on the maintenance of SIB in RS demonstrates that not all cases can be attributed entirely to organic dysfunction, with environmental factors also having an impact (Iwata et al., 1991; Oliver et al., 1986; Roane et al., 2001; Wehmeyer, Bourland & Ingram, 1993).

The main characteristic feature of Lesch Nyhan syndrome (LNS) is severe and intractable SIB, usually in the form of lip and finger biting (Hall et al., 2001). This behaviour is prevalent in almost all cases of LNS and emerges at around the third year of life (Christie et al., 1982). There is a general consensus that SIB in LNS is predominantly biologically driven (Lesch & Nyhan, 1964; Moss et al., 2005). However, a number of researchers have demonstrated the importance of environmental factors, such as social attention, in the maintenance of SIB in LNS (e.g. Anderson, David, Bonnet & Dancis, 1979; Hall et al., 2001).

Finally, in Cornelia de Lange syndrome (CdLS), a number of studies have explored SIB and prevalence estimates range from 17-63.6% (Beck, 1987; Berney et al., 1999; Hyman et al., 2002). The most common forms of self-injurious behaviour observed in individuals with CdLS are biting and scratching (Arron, et al., in press; Gualtieri, 1990; Sarimski, 1997). As is the case for Lesch-Nyhan syndrome, it has been suggested by some that SIB in CdLS is biologically determined (Nyhan, 1972; Carr, 1977; Hyman et al., 2002). However, research findings have demonstrated that SIB in individuals with CdLS is associated with environmental changes. For example, Moss et al. (2005) found that in seven participants with CdLS, some forms of SIB were associated with particular setting events. These findings indicate a need to look beyond the assumption that SIB in genetic syndromes is determined

purely by biological factors and that multiple factors are likely to be at play (Moss et al., 2005). More recently, the interactions between biological and developmental systems and the enviroment have become the focus of research (e.g. Hall et al., 2001).

Environmental determinants of self-injurious behaviour

There is an extensive and robust literature on the *aetiology* of SIB which demonstrates a role for social reinforcement (Hanley, Iwata & McCord, 2003; Oliver, 1995). There is also a wealth of literature that provides evidence for a role for social reinforcement in the *maintenance* of established SIB with evidence from studies using a range of research methods including experimental functional analytic (Iwata et al., 1982; Carr and Durand, 1985), naturalistic observation (Emerson et al., 1996; Lerman and Iwata, 1993) and behavioural intervention studies (Emerson, 2000). Operant theory provides a useful framework for understanding both the development and maintenance of SIB.

The theory of operant conditioning proposes that behaviour occurs based on the consequences that were previously associated with its occurrence (Roane et al., 2001). Carr (1997) proposed that SIB is an operant behaviour, and suggested three ways in which the development and maintenance of SIB can occur: positive, negative and automatic reinforcement. The first (positive reinforcement) suggests that SIB is maintained by the contingent presentation of a stimulus following a behaviour, for example, social attention. The second (negative reinforcement) involves the contingent cessation of an aversive event, such as a demanding task. The third (automatic reinforcement) describes behaviour that occurs because it either produces or alleviates a source of stimulation independent of the social environment. In order to identify the potential reinforcing properties of SIB, Iwata, Dorsey, Slifer, Bauman and Richman (1982/94) developed an experimental method of functional analysis to test Carr's hypotheses.

Assessing the function self-injurious behaviour

Iwata et al. (1982/94) developed an operant methodology which has subsequently become a favoured method for the study of environmental determinants of SIB (Moss et al., 2005). The method was designed to identify functional relations between the occurrence of SIB and specific environmental events. Three test conditions are presented during the functional analysis in which events that occur prior to a behaviour (such as the presence or absence of social attention, aversive stimulation or activities) and events that follow a behaviour (such as

the delivery of attention, escape from tasks, or no stimulation) are varied and associated changes in SIB are measured. Each of the test conditions is designed to test a particular operant reinforcer. Using this method of experimental functional analysis, Iwata, Pace, Dorsey, Zarcone, Vollmer & Smith et al. (1994) carried out a large scale analysis of the reinforcing functions of SIB. They examined data from 152 single subject analyses over an eleven year period and found that social-negative reinforcement (escape from demands or other sources of aversive stimulation) accounted for the largest proportion of the sample (38.1%). Social-positive reinforcement (either attention or access to food or materials) accounted for 25.7% and automatic reinforcement accounted for 25.7%. Multiple controlling variables accounted for 5.3% of the cases. There has since followed a substantial empirical evidence base within the operant learning literature to suggest that social attention has a strong reinforcing effect on SIB (e.g. Iwata et al., 1994; Moss et al., 2005; Oliver, Hall & Murphy, 2005). Exploration of the reinforcing properties of hand mouthing, more specifically have also received attention, with a focus on social and automatic reinforcement.

Environmental determinants of hand mouthing

Several theories have been proposed to explain the aetiology of hand mouthing. A common feature of these theories is that hand mouthing is viewed as a learned response, with a source of reinforcement to which behavioural maintenance is attributed (Goh et al., 1995). For example, researchers have suggested that hand mouthing is maintained by social consequences, in the form of either attention (via positive reinforcement) or escape from a demand (via negative reinforcement) (Baumeister & Forehand, 1973; Hall et al., 2001; Moss et al., 2005; Roane et al., 2001). Attention and escape are social forms of reinforcement, whereas increased (or decreased) sensory stimulation and alleviation of discomfort are non-social forms and referred to as automatic reinforcement (Vaughan & Michael, 1982). Goh et al., 1995 carried out an analysis of the reinforcing properties of hand mouthing. They reported that the prevailing view in the literature during the 1990's emphasised the self-stimulatory (i.e. automatically reinforced) nature of hand mouthing and for this reason, research tended to focus on treatments of hand mouthing. Their review of eight studies that attempted to identify how hand mouthing was maintained found that whilst outcomes were consistent with the view that most hand mouthing is maintained by automatic reinforcement, they were also indicative of social reinforcement functions. Numerous studies have since examined the reinforcing properties of hand mouthing and this paper aims to provide an updated review of the research outcomes and more specifically, whether hand mouthing was

found to be positively reinforced (through social attention), negatively reinforced (through demand escape) or automatically reinforced (through self-stimulation). This review will also provide a brief summary of the research treatments of hand mouthing, as this is covered in detail elsewhere in the literature (e.g. Canella, O'Reilly, & Lancioni, 2006).

Search Criteria

The electronic databases PsycInfo®, Embase® and Medline® were used to carry out a literature search using the search terms in Table 1.

Table 1: Terms employed in the literature search for studies

Search term	Variations
Hand mouthing	Hand biting; hand sucking; self biting; finger sucking; finger biting.
Intellectual disability	Learning disability; intellectual impairment; developmental disorder; mental handicap; mental retardation.

Studies were included in the review if they investigated the assessment and /or treatment of hand mouthing with participants with severe to profound intellectual or multiple disabilities. Only studies conducting experimental functional analysis were selected for further review. Studies were categorised as having conducted a functional analysis if they conducted an experimental manipulation of antecedent or consequent events that were hypothesised to maintain hand mouthing. In addition, only clinical and experimental studies and case studies published between 1995 and 2009 from peer reviewed journals were included. Further hand searches were conducted using the reference sections of the articles identified through the electronic searches to identify a more complete set of papers. Thirty studies were identified and included in this review. Table 2 lists the studies included in the review, grouped according to how the hand-mouthing was found to be maintained. For each study, the table reports the number of participants involved, their gender, ages, level of intellectual disability and topography of SIB (hand mouthing, hand biting, or finger sucking).

Table 2: Studies listed according to maintaining function of hand mouthing, number, level of disability, gender, age and topography of the behaviour

Category/ author	n[1]	Level of disability	Gender	Age[2]	Self-injury
Positive reinforcement					
Goh et al. (1995)[3]	2	Profound	F	25-40	Hand mouthing
Fischer et al. (1997)[3]	1	Profound	F	44	Hand mouthing
O'Reilly et al. (2000)[3]	1	Severe	F	9	Hand mouthing
Lohrmann-O'Rourke and Yurman (2001)[4,5]	1	Severe Multiple	M	6	Hand mouthing
Negative reinforcement					
Golonka et al. (2000)[4]	1	Severe Multiple	M	30	Hand biting
Lohrmann-O'Rourke and Yurman (2001)[4,5]	1	Severe Multiple	M	6	Hand mouthing
McCord, Thompson, and Iwata (2001)$_3$	1	Profound	M	38	Hand biting
Automatic reinforcement					
Derby et al. (1995)[4]	1	Profound	F	3	Hand mouthing
Goh et al. (1995)[3]	10	Profound	M, F	24-68	Hand mouthing
Realon et al. (1995)[4]	1	Profound	F	15	Hand mouthing
Irvin et al. (1996)[4]	3	Profound, Profound multiple	F	31-44	Hand mouthing

Category/ author	n[1]	Level of disability	Gender	Age[2]	Self-injury
Lerman and Iwata (1996)[3]	1	Profound	M	32	Hand mouthing
Turner, Realon, Irvin and Robinson (1996)[4]	3	Profound, Profound multiple	M, F	21-40	Hand mouthing
Ringdahl et al. (1997)[3]	1	Severe	M	5	Hand mouthing
Shore et al. (1997)[3]	2	Profound	F	30-31	Hand mouthing
Lerman et al. (1997)[3]	4	Profound	M, F	25-39	Hand mouthing
Irvin et al. (1998)[3]	2	Profound	F	25 - 41	Hand mouthing
Luiselli (1998)[4]	1	Severe Multiple	M	15	Hand mouthing
Shirley, Iwata & Kahng (1999)[3]	1	Profound	F	26	Hand mouthing
Wallace, Iwata, Zhou & Goff (1999)[3]	1	Profound	F	Un	Hand mouthing
Hanley, Iwata, Thompson & Lindberg (2000)[3]	1	Profound Multiple	M	46	Hand mouthing
Roscoe et al. (1998)[3]	1	Profound	F	35	Hand mouthing
Piazza et al. (2000)[3]	1	Profound	M	17	Hand mouthing
Category/ author	**n[1]**	**Level of disability**	**Gender**	**Age[2]**	**Self-injury**
Zhou et al. (2000)[3]	4	Profound	F	33-51	3 Hand mouthing, 1 Finger sucking
Lohrmann-O'Rourke and Yurman (2001)[4,5]	1	Severe Multiple	M	6	Hand mouthing

Roane et al (2001)[3]	1	Rett syndrome	F	32	Hand mouthing
Lindberg at al. (2003)[3]	1	Profound	F	37	Hand mouthing
Lancioni, Singh, O'Reilly, Oliva, Campodonico & Groenweg (2004)	1	Profound	M	23	Hand mouthing
Lancioni, O'Reilly, Singh, Sigafoos, Oliva, Baccani & Groenweg (2006)	1	Profound	M	12	Finger mouthing
Lancioni, Singh, O'Reilly, Sigafoos, Oliva, Pidala, et al. (2007)[4]	1	Profound	M	13	Hand mouthing
Lancioni, Singh, O'Reilly, Sigafoos, Oliva, Severini, et al. (2007)[4]	1	Profound	F	8	Hand mouthing
Canella-Malone et al. (2008)[4]	1	Profound	M	10	Hand mouthing
Lancioni et al (2008)[4]	1	Profound	F	12	Hand mouthing

Positive reinforcement

There were five individuals from four studies for which hand-mouthing was found to be maintained via positive reinforcement (Fischer et al., 1997; Goh et al., 1995; Lohrmann-O'Rourke & Yurman, 2001; O'Reilly, Lancioni, King, Lally & Dhomhnaill, 2000). For example, O'Reilly et al. (2000) found the hand mouthing behaviour of a nine year old girl to be attention maintained under a specific social context, when her parents were engaged in social interactions with a third person. Outcomes from functional analysis carried out by Goh et al. (1995) with two women, aged 25 and 40 years old found hand mouthing to be positively reinforced. For one woman, hand mouthing was highest when she was gaining attention, suggesting that her behaviour was maintained by positive reinforcement in the form of attention. For the other woman, hand mouthing was highest when she had access to materials, indicating that her behaviour was maintained by access to an object.

Negative reinforcement

There were three individuals from three studies for which hand mouthing was found to have a demand escape function via negative reinforcement (Golonka, Wacker, Berg, Derby, Harding & Peck, 2000; Lohrmann-O'Rourke & Yurman, 2001; McCord, Thomspon & Iwata, 2001). For example, McCord et al. (2001) carried out functional analysis with a 38 year old man, whose hand biting was maintained by avoidance of having to change locations and of certain tasks, for example brushing his teeth. Lorhmann-O'Rourke & Yurman (2001) found that in a 6 year old boy, hand mouthing had multiple functions, occurring in alone time, in a group setting and serving a demand escape function when he was required to do skill building tasks.

Automatic reinforcement

For the remaining 47 participants from 26 studies, hand mouthing was found to be automatically reinforced. The largest scale study was that carried out by Goh et al. (1995) who found that ten out of twelve individuals, both males and females ranging in age from 24-68 years old, showed the highest levels of hand mouthing when they were functionally alone, indicating that the behaviour was automatically reinforced. Similarly Zhou, Goff and Iwata (2000) found the hand mouthing behaviour of four women with profound disabilities to persist in the absence of social stimulation, not varying in the presence of social consequences (attention or escape).

Comparison with previous literature

This review examined 30 studies that had established the reinforcing properties of hand mouthing of 53 individuals with severe to profound intellectual disabilities. For the majority of individuals (47, 88.7%) hand mouthing was found to be automatically reinforced. For five individuals (9.4%), hand

mouthing was found to be positively reinforced and for three individuals (5.7%), it was found to be negatively reinforced. There was one participant (Lohrmann-O'Rourke & Yurman, 2001) for whom hand-mouthing had multiple functions.

Compared with the findings from Iwata et al. (1994) who found that social-negative reinforcement accounted for 38.1%, social-positive reinforcement 25.7% and automatic reinforcement accounted for 25.7% of SIB, the current review suggests that a much higher proportion of hand mouthing behaviour is maintained independently of the social environment. This also reflects the findings from a review of eight studies in the 1990's (Goh et al., 1995) that indicated most hand mouthing to be maintained by automatic reinforcement, with some indicative of social reinforcement functions. The finding that such a high proportion of hand mouthing appears to be maintained by automatic reinforcement has a number of implications for both understanding the aetiology and treatment of hand mouthing as SIB.

Treatment of hand mouthing

Over the last two decades, treatment of SIB in general and of hand mouthing has moved away from using aversive interventions, such as punishment and restraint. Canella et al. (2006) carried out a review of a wide array of interventions that have been used to reduce rates of hand mouthing, including antecedent interventions (such as environmental enrichment and choice), reinforcement-based treatments, sensory stimulation, and multi-component treatments, combining more than one approach. Their review of the literature on treatment of hand mouthing in individuals with severe to profound developmental disabilities resulted in a number of key findings. Of 20 treatment studies published between 1995 and 2004, eighteen reported positive treatment outcomes, with the remaining two studies reporting mixed results. This finding suggests that a variety of different intervention approaches can be successful in reducing hand mouthing. However, the authors urge caution, given that unsuccessful treatment outcomes may not be as widely reported in the literature as successful outcomes.

In addition, the majority of the interventions reviewed were implemented by a researcher (rather than a parent or direct caregiver) and in hospital, institution or segregated classroom settings. This has implications for the practicality of continuing these interventions in home environments or, for example, in school settings, on a longer term basis, where a member of the research team would not be present. Furthermore, it is interesting to note that some interventions are implemented for relatively short periods, with little or no follow up of outcomes (e.g. Derby et al., 1995). In

contrast, other studies have carried out longer term interventions, for example, McCord et al., (2001) carried out baseline and intervention periods spanning up to 120 days.

A further issue is that of pre-treatment assessment. Canella et al. (2006) found that seventeen of the studies they reviewed conducted some form of functional analysis either prior to or during the study to determine the function of hand mouthing. In addition, some studies carried out preference assessment procedures to help inform intervention choices. For example, Piazza et al. (2000) provided one participant who displayed hand mouthing that was hypothesised to be reinforced by either hand or mouth stimulation with a range of stimuli (e.g. a hand massager and a mouth guard) and recorded which stimulus was associated with lowest rate of hand mouthing. The outcome was used to identify whether hand mouthing was reinforced by oral or hand stimulation and the stimulus used as part of the subsequent intervention. Although it appears as if functional analysis and preference assessment procedures are being widely used in the research literature, again, Canella et al. (2006) suggest that in many studies, the potential of these procedures for informing interventions is not being realised. They note that a number of studies report that the results from a functional analysis form the basis of an intervention (e.g. Hanley, Iwata, Thompson & Lindberg, 2000; Roane et al., 2001) but that no explicit link between the two is made. Pelios, Morren, Tesch and Axelrod (1999) carried out a review of the impact of functional analysis methodology on treatment choice for SIB. They concluded that when environmental events that maintain a behaviour are identified, researchers are more like to choose reinforcement-based interventions rather than more aversive interventions. Indeed, previous research has indicated that when the functional characteristics of treatment are matched to those of the behaviour, reinforcement-based procedures can be highly effective (Neef & Iwata, 1994). This emphasises the importance of examining the reinforcing properties of hand mouthing behaviour prior to intervention.

With regard to behaviours that have been found to be automatically reinforced (including hand mouthing), some suggestions have been made to aid in the development of appropriate interventions. LeBlanc, Patel and Carr (2000) reviewed studies that had evaluated assessment strategies for behaviour maintained by automatic reinforcement. They suggest three types of assessment that might lead to enhancement in the development of interventions: non-hypothesis based stimulus preference assessments; hypothesis-based stimulus preference assessments and hypothesis-based treatments that incorporate reinforcement and sensory elements. LeBlanc et al. (2001) recommend that further research should examine whether treatments informed by assessment procedures have a substantial benefit over treatments developed with less assessment data. If this is the case, it would suggest that more focused pre-intervention assessment in

automatically maintained hand mouthing could enhance the effectiveness of interventions. Additionally, a more comprehensive understanding of the aetiology of automatically maintained hand mouthing would assist researchers in developing more tailored, effective interventions.

Aetiology of hand mouthing

Automatic reinforcement is a general term and, arguably, is used to described what the reinforcer is not (i.e. a social variable) and does not necessarily identify a specific reinforcer (Piazza, Adelinis, Hanley, Goh & Delia, 2000). There have been several theories put forward to try to explain how automatic reinforcement might maintain hand mouthing behaviour. Guess (1966) examined stereotyped behaviour in individuals with and without sensory deficits, and concluded that hand mouthing provided increased stimulation to those with sensory deficits, such as impaired sight and mobility. This would suggest that hand mouthing is maintained by automatic positive reinforcement. In contrast, Baumeister and Forehand (1973) proposed that hand mouthing may act to decrease stimulation by blocking the sensory input produced by noisy or crowded environments, suggesting that automatic negative reinforcement is occurring. Rast and Jack (1992) put forward a third explanation, that hand mouthing becomes a 'vicious cycle' where at first, hand mouthing causes discomfort, through the hands being dry or chapped. Hand mouthing may then act to provide temporary alleviation of the hands when saliva comes into contact with the chapped skin. However, continual hand mouthing can then lead to further skin damage, as enzymes in the saliva act to break down the tissue, which in turn keeps the cycle of behaviour maintained.

These three theories all view hand mouthing as a learned response, however, if this was the case, it would suggest that all individuals with a profound level of intellectual disability are equally likely to develop SIB and more specifically hand mouthing behaviour. As previously discussed, individuals from a number of genetic syndrome groups have been found to be at a higher risk for SIB, and more specifically hand mouthing behaviours (Arron et al., in press; Berney, Ireland, & Burn, 1999; Christie et al., 1982; Largo & Schinzel, 1985; Hall et al., 2001; Lesch & Nyhan, 1964; Moss et al., 2005; Oliver et al., 1986; Steinhausen, et al., 2002; Symons et al., 2003) suggesting that a straightforward behavioural explanation is not adequate.

Gene /environment interaction

Examining genetic syndromes in which the prevalence of hand mouthing behaviour is elevated allows for exploration of the role of biological and environmental determinants. If operant learning theory alone could explain hand mouthing behaviour, then we would expect to see no differences in prevalence across syndrome groups, as environmental influences could be presumed to be randomly

distributed across groups (Arron et al., in press). There has been research examining the correlates of SIB in different syndrome groups, suggesting that lower affect, and higher levels of overactivity, impulsivity and repetitive behaviour are all associated with higher levels of SIB (Arron et al., in press; Bodfish, Crawford, Powell, Parker, Golden & Lewis, 1995; Marr, this volume; Marston, Perry & Roy, 1997; Petty & Oliver 2005). However, these findings are consistent with the identified correlates of SIB in individuals with intellectual disabilities in general.

To date, relatively little exploration of automatic reinforcement in hand mouthing specifically has taken place. In fact, automatic reinforcement has been noted to be 'virtually impossible to sever the behaviour from its product, and thus impossible to manipulate the variable of which the behaviour is considered a function' (Vaughan & Michael, 1982; pg. 224). One suggestion has been to provide access to sources of stimulation similar to that produced by hand mouthing to see which source is an effective substitute for hand mouthing contact (Green & Freed, 1993). For example, relative preference for mouth-object contact may suggest stimulation of the mouth as a predominant reinforcer, where a preference for hand-object contact may suggest that stimulation of the hand is the predominant reinforcer. To investigate this idea, Goh et al. 1995, carried out experiments with five individuals who had previously shown hand mouthing maintained by automatic reinforcement. They determined that for all subjects there was a preference for hand stimulation. This outcome had important implications for treatment, as alternative forms of stimulation could be provided.

Some researchers have suggested that pain or discomfort may be a contributing factor to the development and maintenance of SIB in some individuals (Moss et al. 2005; Symons, 2002). Indeed, this has been observed in some studies examining reinforcement of hand mouthing. For example, Lohrmann-O'Rourke and Yurman (2001) found functional analysis with a 6 year old boy to reveal that hand mouthing was more likely to occur when he was suffering from a sinus infection. This finding is supportive of a link between facial discomfort and hand mouthing. Furthermore, over the last decade there has been an emerging literature to support a link between gastroesophageal reflux (GER) and increased SIB, including hand mouthing. Luzzani, Macchini, Valade, Milani and Selicorni (2003) looked at GER in individuals with CdLS and found that the prevalence of self-injury between those with and without GER to be widely different (although this did not reach statistical significance). They also report that SIB improved after treatment of GER, which strongly supports a degree of correlation between the two. Moss, et al., 2005 also report a case where an individual with CdLS stopped exhibiting SIB completely following an operation to correct painful GER, providing further support for this link. Further exploration of this association is important, as it has direct implications for treatment.

Conclusions

This paper reviewed and discussed studies that investigated the reinforcing properties of hand mouthing via experimental functional analysis. Based on the research available, it appears evident that in the majority of individuals, hand mouthing is maintained by automatic reinforcement, that is, independently of the social environment. A brief review of the outcomes from treatment studies suggests that although the majority of outcomes are reported to be positive, caution is warranted as it is possible that many unsuccessful interventions are not reported in the literature. The implications of pre-intervention functional analysis for informing interventions suggest that if the reinforcing properties of hand mouthing are considered when developing interventions, the outcome may be more successful.

Discussion of the literature on automatic reinforcement of hand mouthing behaviour reveals that the reinforcement may have a sensory element (either positive or negative), may be maintained by a vicious cycle of soothing damaged tissue, or may be related to pain, such as that produced by gastro-oesophageal reflux. In the broader context, it is important to consider the ethical implications of reducing hand mouthing behaviour in people with severe and profound intellectual and developmental disabilities. As discussed previously, hand mouthing can have serious social consequences, limiting interaction and integration with others due to the sights and smells it may produce. It is important, therefore to consider the purpose of attempting to reduce hand mouthing, if the behaviour is an issue of social acceptance, yet causing no direct distress to the individual. Perhaps more importantly where automatic reinforcement is found to maintaining hand mouthing, a closer look should be taken at the possible causes of hand mouthing, particularly where there is a possibility that it may be maintained by physical discomfort.

References

Anderson, L. T., David, R., Bonnet, K., & Dancis, J. (1979). Passive avoidance learning in Lesch-Nyhan disease: effect of 1-desamino-8-arginine-vasopressin. *Life Sciences, 24*, 905-910.

Arron, K., Oliver, C., Berg, K., Moss, J., & Burbidge, C. (in press). Delineation of behavioural phenotypes in genetic syndromes: 2. Prevalence, phenomenology and correlates of self-injurious and aggressive behaviour. *Journal of Autism and Developmental Disorders.*

Ball, T. S., Campbell, R., & Barkmeyer, R. (1980). Air splints applied to control self-injurious finger sucking in profoundly retarded individuals. *Journal of Behavior Therapy & Experimental Psychiatry, 11,* 267-271.

Baumeister, A. A., & Forehand, R. (1973). Stereotyped acts. In N. R. Ellis (Ed.), *International review of research in mental retardation* (pp. 55-96). New York: Academic Press.

Beck, B. (1987). Psycho-social assessment of 36 de Lange patients. *Journal of Mental Deficiency Research, 31*, 251-257.

Berkson, G., Tupa, M., & Sherman, L. (2001). Early development of stereotyped and self-injurious behaviors: I. Incidence. *American Journal on Mental Retardation, 106*, 539–547.

Berney, T. P., Ireland M., & Burn, J. (1999). Behavioural phenotype of Cornelia de Lange syndrome. *Archives of Disease in Childhood, 81*, 333-336.

Bodfish, J. W., Crawford, T. W., Powell, S. B., Parker, D. E., Golden, R. N., & Lewis, M. H. (1995). Compulsions in Adults with Mental-Retardation - Prevalence, Phenomenology, and Comorbidity with Stereotypy and Self-Injury. *American Journal on Mental Retardation, 100,* 183-192.

Canella, H. I., O'Reilly, M. F., & Lancioni, G. E. (2005). Treatment of hand mouthing in individuals with severe to profound developmental disabilities: A review of the literature. *Research in Developmental Disabilities, 27,* 529-544.

Canella-Malone, H. I., O'Reilly, M. F., Sigafoos, J., & Chan, J. M. (2008). Combined curricular intervention with brief hands down to decrease hand mouthing and the use of arm splints for a young boy with profound disabilities. *Education and Training in Developmental Disabilities, 43* (3), 360-366.

Carr, E. G. (1977). The motivation of self-injurious behaviour: a review of some hypotheses. *Psychological Bulletin, 84,* 800-816.

Carr, E.G., & Durand, V.M. (1985). Reducing behavior problems through functional communication training. *Journal of Applied Behavior Analysis, 18*, 111-126.

Christie, R., Bay, C., Kaufman, I.A., Bakay, B., Borden, M., & Nyhan, W.L. (1982). Lesch-Nyhan disease: Clinical experience with nineteen patients. *Developmental Medicine and Child Neurology, 24,* 293-309.

Cooper, S. A., Smiley, E., Allan, L. M., Jackson, A., Finlayson, J., Mantry, D., & Morrison, J. (2009). Adults with intellectual disabilities: prevalence, incidence and remission of self-injurious behaviour, and related factors. *Journal of Intellectual Disability Research, 53* (3), 200-16.

Derby, K. M., Wacker, D. P., Andelman, M., Berg, W., Drew, J., Asmus, J., et al. (1995). Two measures of preference during forced-choice assessments. *Journal of Applied Behavior Analysis, 28,* 345-346.

Emerson, E., Reeves, D., Thompson, S., Henderson, D., Roberston, J. & Howard, D. (1996*).* Time-based lag sequential analysis and the functional assessment of challenging behaviour. *Journal of Intellectual Disability Research, 40,* 260-274.

Emerson, E. (2000). *Challenging Behaviour. Analysis and Intervention in People with Intellectual Disabilities* (2nd ed.). Cambridge University Press: Cambridge.

Emerson, E., Kiernan, C., Alborz, A., Reeves, D., Mason, H., Swarbrick, R., Mason, L., & Hatton, C. (2001). Predicting the Persistence of Severe Self-Injurious Behavior. *Research in Developmental Disabilities*, *22* (1), 67-75.

Fischer, S. M., Iwata, B. A., & Mazaleski, J. L. (1997). Noncontingent deleiver of arbitrary reinforcers as treatment for self-injurious behaviour. *Journal of Applied Behavior Analysis, 30,* 239-249.

Goh, H. L., Iwata, B. A., Shore, B. A., DeLeon, I. G., Lerman, D. C., Ulrich, S. M., & Smith, R. G. (1995). An analysis of the reinforcing properties of hand mouthing. *Journal of Applied Behavior Analysis, 28* (3), 269-83.

Golonka, Z., Wacker, D., Berg, W., Derby, K. M., Harding, J., & Peck, S. (2000). Effects of escape to alone versus escape to enriched environments on adaptive and aberrant behavior. *Journal of Applied Behavior Analysis, 33,* 243-246.

Gualtieri, C. T. (1990). *Neuropsychiatry and Behavioral Pharmacology*: Springer Verlag: Berlin.

Guess, D. (1966). The influence of visual and ambulation restrictions on stereotyped behavior. *American Journal of Mental Deficiency, 70,* 542-547.

Hall, S. S., Oliver, C., & Murphy, G. (2001). Self-injurious behaviour in young children with Lesch-Nyhan Syndrome. *Developmental Medicine and Child Neurology, 43* (11), 745-749.

Hanley, G. P., Iwata, B. A., Thompson, R. H., & Lindberg, J. S. (2000). A component analysis of "stereotypy as reinforcement" for alternative behavior. *Journal of Applied Behavior Analysis, 33,* 285-297.

Hanley, G. P., Iwata, B. A., & McCord, B. E. (2003). Functional analysis of problem behavior: a review. *Journal of Applied Behavior Analysis, 36* (2), 147-85.

Hyman, P., Oliver, C., & Hall, S. (2002). Self-Injurious behaviour, self-restraint, and compulsive behaviours in Cornelia de Lange syndrome. *American Journal on Mental Retardation, 107,* (2), 146-154.

Irvin, D. S., Realon, R. E., Hartley, J. R., Phillips, J. F., Bradley, F., & Daly, M. (1996). The treatment of self-injurious hand mouthing by using a multi-component intervention with individuals positioned in a small group. *Journal of Developmental and Physical Disabilities, 8,* 43-59.

Irvin, D. S., Thompson, T. J., Turner, W. D., & Williams, D. E. (1998). Utilizing increased response effort to reduce chronic hand mouthing. *Journal of Applied Behavior Analysis, 31,* 375-385.

Iwata, B.A., Dorsey, M.F., Slifer, K.J., Bauman, K.E., & Richman, G.S. (1982). Toward a functional analysis of self-injury. *Analysis and Intervention in Developmental Disorders, 2*, 3-20.

Iwata, B., Pace, G. M., Willis, K. D., Gamache, T. B., & Hyman, S. L. (1986). Operant studies of self-injurious hand biting in the Rett syndrome. *American Journal of Medical Genetics, 24,* 157-166.

Iwata, B. A., Pace, G. M., Dorsey, M. F., Zarcone, J. R., Vollmer, T. R., Smith, R. G., Rodgers, T. A., Lerman, D. C., Shore, B. A., & Mazaleski, J. L. (1994). The functions of self-injurious behavior: an experimental-epidemiological analysis. *Journal of Applied Behavior Analysis, 27* (2), 215-40.

Lancioni, G. E., Singh, N. N., O'Reilly, M. F., Oliva, D., Campodonico, F., & Groeneweg, J. (2004). Impact of favorite stimuli on the behavior of a person with multiple disabilities while using a treadmill. *Journal of Visual Impairment and Blindness, 98,* 304-309.

Lancioni, G. E., O'Reilly, M. F., Singh, N. N., Sigafoos, J., Oliva, D., Baccani, S., & Groeneweg, J. (2006). Microswitch clusters promote adaptive responses and reduce finger mouthing in a boy with multiple disabilities. *Behavior Modification, 30,* 892-900.

Lancioni, G. E., Singh, N. N., O'Reilly, M. F., Sigafoos, J., Oliva, D., Pidala, S., Piazzolla, G., & Bosco, A. (2007). Promoting adaptive foot movements and reducing hand mouthing and eye poking in a boy with multiple disabilities through microswitch technology. *Cognitive Behavior Therapy, 36,* 85-90.

Lancioni, G. E., Singh, N. N., O'Reilly, M. F., Sigafoos, J., Oliva, D., Severini, L., Smaldone, A., & Tamma, M. (2007). Microswitch technology to promote adaptive responses and reduce mouthing in two children with multiple disabilities. *Journal of Visual Impairment and Blindness, 101,* 628-636.

Lancioni, G. E., Singh, N. N., O'Reilly, M. F., Sigafoos, J., Didden, R., Oliva, D., Cingolani, E. (2008). A girl with multiple disabilities increases object manipulation and reduces hand mouthing through a microswitch-based program. *Clinical Case Studies, 7,* 238-249.

Largo, R. H., & Schinzel, A. (1985). Developmental and Behavioral Disturbances in 13 Boys with Fragile X-Syndrome. *European Journal of Pediatrics, 143* (4), 269-275.

LeBlanc, L. A., Patel, M. R., & Carr, J. E. (2000). Recent advances in the assessment of aberrant behavior maintained by automatic reinforcement in individuals with developmental disabilities. *Journal of Behavior Therapy and Experimental Psychiatry, 31,* 137-154.

Lerman, D.C., & Iwata, B. A. (1993). Descriptive and experimental analyses of variables maintaining self-injurious behaviour. *Journal of Applied Behavior Analysis, 26*, 293-319.

Lerman, D. C., & Iwata, B. A. (1996). A methodology for distinguishing between extinction and punishment effects associated with response blocking. *Journal of Applied Behavior Analysis, 29* (2) 231-233.

Lerman, D. C., Iwata, B. A., Shore, B. A., & DeLeon, I. G. (1997). Effects of intermittent punishment on self-injurious behavior: An evaluation of schedule thinning. *Journal of Applied Behavior Analysis, 30,* 187-201.

Lesch, M., & Nyhan, W. L. (1964). A familial disorder or uric acid metabolism and central nervous system function. *American Journal of Medicine, 36,* 561-570.

Lindberg, J. S., Iwata, B. A., Roscoe, E. M., Worsdell, A. S., & Hanley, G. P. (2003). Treatment efficacy of noncontingent reinforcement during brief and extended application. *Journal of Applied Behavior Analysis, 36,* 1-19.

Lohrmann-O'Rourke, S., & Yurman, B. (2001). Naturalistic assessment of an intervention for mouthing behaviour influenced by establishing operations. *Journal of Positive Behavioral Interventions, 2,* 19-27.

Luiselli, J. K. (1998). Treatment of self-injurious hand-mouthing in a child with multiple disabilities. *Journal of Developmental and Physical Disabilities, 10,* 167-174.

Luzzani, S., Macchini, F., Valade, A., Milani, D., & Selicorni, A. (2003). Gastroesophageal reflux and Cornelia de Lange syndrome: typical and atypical symptoms. *American Journal of Medical Genetics, 119a,* 283-287.

Marr, A. (2009). Delineation of the behavioural phenotype of 1p36 deletion syndrome: Prevalence, correlates and experimental functional analysis of self-injurious and aggressive behaviour. Unpublished D. Clin. Psych thesis. University of Birmingham, Birmingham, England.

Marston, G. M., Perry, D. W., & Roy, A. (1997). Manifestations of depression in people with intellectual disability. *Journal of Intellectual Disability Research, 41*, 476-480.

McCord, B. E., Thomson, R. J., & Iwata, B. A. (2001). Functional analysis and treatments of self-injury associated with transitions. *Journal of Applied Behavior Analysis, 34,* 195-210.

McClure, J. T., Moss, R. A., McPeters, J. W., & Kirkpartrick, M. A. (1986). Reduction of hand mouthing by a boy with profound mental retardation. *Mental Retardation, 24,* 219-222.

Moss, J., Oliver, C., Hall, S., Arron, K., Sloneem, J., & Petty, J. (2005*)* The association between environmental events and self-injurious behaviour in Cornelia de Lange syndrome. *Journal of Intellectual Disability Research, 40*, 69-77.

Murphy, G., & Wilson, B. (1985). *Self-injurious Behaviour*. British Institute of Learning Disabilities, Kidderminster.

Neef, N. A., & Iwata, B. A. (1994). Current research on functional analysis methodologies: An introduction. *Journal of Applied Behavior Analysis, 27,* 211-214.

Nyhan, W. L. (1972). Behavioral phenotypes in organic genetic disease: presidential address to the Society for Pediatric Research. *Pediatric Research, 6,* 1-9.

Oliver, C., Murphy, G. H., & Corbett, J. A. (1987). Self-injurious behaviour in people with mental handicap: a total population study. *Journal of Mental Deficiency Research, 31*, (2), 147-62.

Oliver, C., Murphy, G. H., Crayton, L., & Corbett, J. (1986). Self-injurious-behavior in Rett syndrome: Interactions between features of Rett syndrome and operant conditioning. *Journal of Autism and Developmental Disorders, 23,* 91-109.

Oliver, C. (1995). Annotation: Self-injurious behaviour in children with learning disabilities: Recent advances in assessment and intervention. *Journal of Child Psychology and Psychiatry, 36*, 909-927.

Oliver, C., Hall, S. & Murphy, G. (2005). The early development of self-injurious behaviour: Evaluating the role of social reinforcement. *Journal of Intellectual Disability Research, 49*, 591-599.

O'Reilly, M. F., Lancioni, G. E., King, L., Lally, G., & Dhomhnaill, O. N. (2000). Using brief assessments to evaluate aberrant behaviour maintained by attention. *Journal of Applied Behavior Analysis, 33,* 109-112.

Pelios, L., Morren, J., & Axelrod, S. (1999). The impact of functional analysis methodology on treatment choice for self-injurious and aggressive behavior. *Journal of Applied Behavior Analysis, 32* (2), 185- 195.

Petty, J., & Oliver, C. (2005). Self-injurious behaviour in people with intellectual disability. *Current Opinion in Psychiatry, 18*, 484-489.

Piazza, C. C., Adelinis, J. D., Hanley, G. P., Goh, H. L., & Delia, M. D. (2000). An evaluation of the effects of matched stimuli on behaviors maintained by automatic reinforcement. *Journal of Applied Behavior Analysis, 33,* 1-27.

Rast, J. & Jack, S. (1992). Mouthing. In E. A. Konarski, J.E. Favell & J. E. Favell (Eds.), *Manual for the assessment and treatment of the behaviour disorders of people with mental retardation* (pp. 1-11). Morganton, NC: Western Carolina Center Foundation.

Realon, R. E., Favell, J. E., & Cacace, S. (1995). An economical, humane, and effective method for short-term suppression of hand mouthing. *Behavioural Interventions, 10,* 141-147.

Ringdahl, J. E., Vollmer, T. R., Marcus, B. E., & Roane, H. S. (1997). An analogue evaluation of environmental enrichment: The role of stimulus preference. *Journal of Applied Behavior Analysis, 30,* 203-216.

Roane, H. S., Piazza, C. C., Sgro, G. M., Volkert, V. M., & Anderson, C. M. (2001). Analysis of aberrant behaviour associated with Rett syndrome. *Disability and Rehabilitation, 23,* 139-148.

Rojahn, J. (1986). Self-injurious and stereotypic behaviour of noninstitutionalised mentally retarded people: prevalence and classification. *American Journal on Mental Deficiency, 91,* 268-276.

Roscoe, E. M., Iwata, B. A., & Goh, H. L. (1998). A comparison of noncontingent reinforcement and sensory extinction as treatments for self-injurious behaviour. *Journal of Applied Behavior Analysis, 31,* 635- 646.

Salovita, T. (2000). The structure and correlates of self-injurious behaviour in an institutional setting. *Research in Developmental Disabilities, 21,* 501-511.

Sansom, D., Krishnan, V. H. R., Corbett, J., & Kerr. (1993). Emotional and behavioural aspects of Rett syndrome. *Developmental Medicine and Child Neurology, 35,* 340-345.

Sarimski, K. (1997). Communication, social-emotional development and parenting stress in Cornelia-de-Lange syndrome. *Journal of Intellectual Disability Research, 41,* 70-75.

Shirley, M. J., Iwata, B. A., & Kahng, S. (1999). False-positive maintenance of self-injurious behavior by access to tangible reinforcers. *Journal of Applied Behavior Analysis, 32,* 201-204.

Shore, B. A., Iwata, B. A., DeLeon, I. G., Kahng, S., & Smith, R. G. (1997). An analysis of reinforce substitutability using object manipulation and self-injury and competing responses. *Journal of Applied Behavior Analysis, 30,* 21-41.

Steinhausen, H. C., von Gontard, A., Spohr, H. L., Hauffa, B. P., Eiholzer, U., Backes, M., et al. (2002). Behavioral phenotypes in four mental retardation syndromes: Fetal alcohol syndrome, Prader-Willi syndrome, fragile X syndrome, and tuberosis sclerosis. *American Journal of Medical Genetics, 111* (4), 381-387.

Symons, F. J., Clark, R. D., Hatton, D. D., Skinner, M., & Bailey, D. B. (2003). Self-injurious behavior in young boys with fragile X syndrome. *American Journal of Medical Genetics Part A*, *118a*, 115-121.

Turner, W. D., Realon, R. E., Irvin, D., & Robinson, E, (1996). The effects of implementing program consequences with a group of individuals who engaged in sensory maintained hand mouthing. *Research in Developmental Disabilities, 17,* 311-330.

Vaughan, M. E., & Michael, J. L. (1982). Automatic reinforcement: An important but ignored concept. *Behaviorism, 10,* 217-227.

Wallace, M. D., Iwata, B. A. Zhou, L., & Goff, G. A. (1999). Rapid assessment of the effects of restraint on self-injury and adaptive behavior. *Journal of Applied Behavior Analysis, 32* (4), 525-8.

Wehmeyer, W., Bourland, G., & Ingram, D. (1993). An analogue assessment of hand stereotypies in two cases of Rett syndrome. *Journal of Intellectual Disabilities Research, 37,* 95-102.
Zhou, L., Goff, G. A., & Iwata, B. A. (2000). Effects of increased response effort on self-injury and object manipulation as competing responses. *Journal of Applied Behavior Analysis, 33,* 29-40.

Empirical Paper

Delineation of the behavioural phenotype of 1p36 deletion syndrome: Prevalence, correlates and experimental functional analysis of self-injurious and aggressive behaviour.

Abstract

Background: Studies of 1p36 deletion syndrome have focussed on physical characteristics with limited exploration of the behavioural phenotype. When behavioural features have been reported, self-injury and aggression are noted. In this study we describe these behaviours and investigate aetiology.

Methods: We compared the prevalence of self-injurious and aggressive behaviour in 1p36 deletion syndrome (n=23) in comparison with three matched syndrome groups: Angelman (n=21); Cri du Chat (n=23) and Cornelia de Lange (n=23) syndromes. Carers completed questionnaires regarding self-injury, physical aggression, mood, autism spectrum disorder, hyperactivity and repetitive behaviour. Experimental functional analysis was carried out with six children

Results: Fourteen (60.9%) participants in the 1p36 deletion syndrome group showed self injury and twelve (52.2%) showed physical aggression, with self biting found to be the most common topography of self-injury. Self-injurious behaviour was associated with overactivity and stereotyped behaviour and aggression was associated with impulsivity and repetitive behaviour. Behavioural data confirmed high levels of hand mouthing and for three participants there was evidence of attention maintained function of self injury.

Discussion: The findings are consistent with previous research which has suggested self-injury to be common in 1p36 deletion syndromes. The implications for treatment of self-injury are discussed within the context of shaping precursor behaviours to have a communicative function prior to the development of self-injurious behaviour.

Introduction

Behavioural phenotypes have been described for a number of rare genetic disorders, for example, Down, Angelman, Prader-Willi, Cornelia de Lange, Cri du Chat, Fragile X and Williams syndromes (see respectively: Chapman & Hesketh, 2000; Dykens & Kasari, 1997; Horsler & Oliver, 2006; Hyman, Oliver & Hall, 2002; Collins & Cornish, 2002; Kau, Reider, Payne, Meyer & Freund, 2000 and Udwin & Yule, 1991). There is currently increasing recognition of possible behavioural phenotypes for microdeletion syndromes, which are rare and not frequently mentioned in the literature (e.g. Fisch, Battaglia,Parrini, Youngblom & Simensen, 2008). A microdeletion is the loss of portion of a chromosome that is so small that its absence is not apparent on ordinary examination and detection requires techniques such as fluorescence in situ hybridisation (FISH) to detect and localise the presence or absence of specific DNA sequences on chromosomes (Slavotinek, Shaffer & Shapira, 1999). One such syndrome which has been recognised as a distinct chromosome deletion syndrome is 1p36 deletion syndrome.

1p36 deletion syndrome

1p36 deletion syndrome has an estimated incidence of 1 in 5,000 to 1 in 10,000 live births with a 2:1 female to male ratio (Shapira, McCaskill, Northrup, Spikes, Elder & Sutton, 1997; Shaffer and Lupski, 2000). It is caused by a deletion of chromosome band 1p36, resulting in global developmental delay and intellectual disability and numerous physical disabilities and medical disorders (Shapira et al., 1997; Heilstedt et al., 2003). Notable characteristics are: cardiovascular problems, hearing and sight loss, epilepsy and feeding difficulties (Heilstedt et al., 2003). The syndrome is also characterised by distinctive facial features, low muscle tone and growth abnormalities. Research to date has tended to focus on the physical characteristics of the syndrome (e.g. Heilstedt et al., 2003; Slavotinek, Shaffer & Shapira, 1999). Where the behavioural phenotype of 1p36 deletion syndrome has been studied, findings from clinical examinations have tended to describe behaviours rather than employ behavioural assessments with known psychometric properties. For example, Knight-Jones, Knight, Heussler, Regan, Flint & Martin (2000) describe case reports on four children with 1p36 deletion sydrome and report that two children showed self-injury (sucking fingers excessively and finger biting). Shapira et al. (1997) describe findings from clinical examinations of thirteen patients as well as parental reports from a checklist based on the previous literature. They found "abusive behaviour" to be reported in seven people. Reish, Berry and Hirsch (1995) carried out clinical examinations on five patients ranging from 3-47 years-old and found all to exhibit some level of "self-abusive" behaviour. Battaglia (2005) reviewed a number of case studies and noted common behavioural features including self injurious behaviour

(hand/wrist biting; hand chewing; head banging; pinching her/himself; scratching the peroneal region), temper outbursts, banging or throwing objects, striking people and episodes of violent physical activity. Anecdotal reports from parents, collated in a report by the syndrome support group UNIQUE, describe children and young people with 1p36 deletion syndrome as very affectionate and sociable.

Self-Injury

Self-injurious behaviour (SIB) can have a significant impact on the quality of life of an individual and their family and remains a challenge to services. Recent literature has focused on exploring the prevalence, cause and treatments of SIB (e.g. Petty & Oliver, 2005; Cooper, Smiley, Allan, Jackson, Finlayson, Mantry & Morrison, 2009). There are two broad domains of research literature that have explored the aetiology of SIB in individuals with intellectual disability. The first demonstrates that SIB can be learned via operant reinforcement (Hanley, Iwata & McCord, 2003; Oliver, 1995) providing evidence for environmental determinants of SIB. The second places emphasis on potential biological determinants of SIB implied by the association between SIB and greater degree of learning disability, the presence of stereotyped, compulsive and impulsive behaviours, autism spectrum disorders and genetic syndromes (Bodfish, Crawford, Powell, Parker, Golden & Lewis, 1995; Brylewski & Wiggs, 1999; Deb, Thomas & Bright, 2001; McClintock, Oliver & Hall 2003; Powell, Bodfish, Parker, Crawford & Lewis, 1996; Rojahn, Matson, Naglieri & Mayville, 2004). Exploring SIB in genetic syndromes, where the prevalence of SIB is noted to be higher than in the wider population of people with intellectual disability, is a useful strategy for exploring the difference between these two perspectives. A number of studies have highlighted that individuals with particular genetic sydromes are at risk for developing SIB, e.g. Cornelia de Lange syndrome; Cri du Chat syndrome and Prader-Willi syndrome (Clarke et al., 1996; Collins & Cornish, 2002; Oliver, Arron & Sloneem, 2008). However, there may be common aetiological pathways as certain behaviours predictor the presence of SIB across a number of different syndrome groups. For example, high levels of overactivity, impulsivity, repetitive and stereotyped behaviours have been found to be associated with self-injury in individuals with Angelman, Cri du Chat, Cornelia de Lange, Fragile X, Prader-Willi, Lowe and Smith Magenis Syndromes. (Arron, Oliver, Berg, Moss and Burbidge, in press; Bradley, Summers, Wood and Bryson, 2004; Marston, Perry and Roy, 1997; Oliver et al. 2008; Petty & Oliver, 2005; Rojahn, Matson, Naglieri and Mayville, 2004).

Comparatively less attention has been paid to aggression than self-injury in syndrome groups. Research literature examining aggressive behaviour has explored the prevalence of aggression in rare syndromes, for example, Angelman (Strachan, Shaw, Burrow, Horsler, Allen & Oliver, 2009)

and Prader-Willi syndromes (Woodcock, Oliver & Humphreys, 2009). Again, there is evidence to show that impulsive and repetitive behaviours are predictors of aggression among individuals from different syndrome groups (Arron, et al., in press; Clarke et al. 2002).

Although a high prevalence of self-injury and aggression has been found in some syndrome groups, there is evidence for the importance of environmental factors in the maintenance of SIB (Arron, Oliver, Hall, Sloneem, Forman & McClintock, 2006; Hall, Oliver & Murphy, 2001; Moss, Oliver, Hall, Arron, Sloneem, & Petty, 2005; Taylor and Oliver, 2008, Strachan et al., 2009). There is substantial evidence within the operant literature to suggest that social reinforcement influences self-injurious and aggressive behaviours (e.g. Iwata, Dorsey, Slifer, Bauman & Richmanm, 1982; Lovaas, Freitag, Gold & Kassorla, 1965; Oliver, Hall & Murphy, 2005). Given that research has indicated this association between genetic sydromes and self-injurious and aggresive behaviour, it is an important behaviour to explore in 1p36 deletion syndrome with regard to prevalence and phenomenology and also to explore potential operant reinforcement. A matched comparison group design and the use of experimental functional analysis respectively can address these questions.

There are two broad aims to the present study: to further define the behavioural phenotype of 1p36 deletion syndrome with regard to self-injury and aggression and to examine the potential environmental determinants of these behaviours.

Study one

Aims of study one

The aim of the first study is to examine the prevalence of self injury and physical aggression in 1p36 deletion syndrome and compare the prevalence and phenomenology to that seen in Angelman, Cri du Chat and Cornelia de Lange syndromes, syndromes in which the distribution of degree of intellectual disability is broadly comparable. In addition, the study aimed to evaluate correlates of self-injury and physical aggression in the 1p36 deletion syndrome group specifically the presence of autism spectrum behaviours, hyperactivity and repetitive behaviours.[3]

Methods

Participants[4]

Individuals with 1p36 deletion syndrome were recruited via Unique, which supports rare syndrome groups. Overall, 54 individuals identified with 1p36 deletion syndrome were contacted for participation in the study. Information regarding the diagnosis of 1p36 deletion syndrome was obtained. Participants were excluded from the study if they did not have a diagnosis from a Clinical Geneticist, General Practitioner, Neurologist, Psychiatrist or Paediatrician or if more than 25% of information was missing from any questionnaire. After excluding participants, 23 individuals with 1p36 deletion syndrome were included in the study an inclusion rate of 42.6%. Matched participants with Angelman, Cornelia de Lange, Cri du Chat syndromes were drawn from an existing database (Moss, Oliver, Arron, Burbidge & Berg, 2008). Participants were matched on self help skills (from the Wessex Scale: Kushlick, Blunden & Cox, 1973) and verbal ability (from parental report). Full details of recruitment procedure and return rates for the matched participant syndrome groups are given in Moss et al., 2008. Participant details for the whole sample, broken down by group, are shown in Table 3. Across the four groups there were 90 participants with a mean age of 10.35 years (standard deviation, 7.96 years). There were 59 (66%) females in the sample with 42 (47%) participants verbal, 31 (34%) mobile and 17 (19%) able[5]. 65 (72%) of the participants had normal hearing and 56 (62%) had normal vision. The mean age, standard deviation, and details of gender, vision, hearing, speech, mobility and level of ability within the four syndrome groups can be found in Table 3.

Table 3: Frequency and percentage of females, mean age (standard deviation), frequency and percentage of participants with normal vision and hearing and who were verbal, mobile and able in each syndrome group and post hoc contrasts for vision and hearing (AS = Angelman Syndrome, CdLS = Cornelia de Lange Syndrome, CDCS = Cri du Chat Syndrome, 1p36 = 1p36 Syndrome)

		Syndrome group				Chi –square / ANOVA			Post Hoc <.01
		1p36	AS	CdCS	CdLS	df	χ /F*	P value	
N		23	21	23	23				
Age[a]	Mean (SD)	7.29 (4.19)	11.64 (10.46)	11.28 (8.09)	11.29 (7.75)	3	1.56*	.21	
	Range	1.05-16.04	1.98-45.08	1.59-33.06	1.89-29.22				
Gender	Female	16	12	15	16	3	.99	.80	
	(%)	(69.6)	(57.1)	(65.2)	(69.6)				
Self help[b]	Partly able/able[c]	4	2	5	6	3	2.14	.55	
	(%)	(17.4)	(9.5)	(21.7)	(26.1)				
Mobility[b]	Fully mobile	10	4	11	6	3	6.36	.10	
	(%)	(43.5)	(19)	(47.8)	(26.1)				
Vision[b]	Normal	8	18	20	10	3	20.75	<.001	AS, CDCS>1p36
	(%)	(34.8)	(85.7)	(87)	(43.5)				
Hearing[b]	Normal	14	21	19	11	3	17.23	<.001	AS>CdLS
	(%)	(60.9)	(100)	(82.6)	(47.8)				
Speech	Verbal	9	6	12	15	3	6.75	.08	
	(%)	(39.1)	(28.6)	(52.2)	(65.2)				

[a] In years (decimal)
[b] Data derived from the Wessex questionnaire (Kushlick et al., 1973)
[c] Based on the ability to feed, dress and wash independently or with help

Measures

Seven questionnaires[6] were sent to carers, these were: a demographic questionnaire, the Wessex Scale (Kushlick, Blunden & Cox, 1973), the Challenging Behaviour Questionnaire (CBQ; Hyman, Oliver & Hall, 2002) , the Mood, Interest and Pleasure Questionnaire Short-form (MIPQ-S; Ross & Oliver, 2003; Ross et al., 2008), the Social Communication Questionnaire- Lifetime (SCQ-L; Rutter Baily & Lord, 2003), The Activity Questionnaire (TAQ; Burbidge & Oliver, 2008), The Repetitive Behaviour Questionnaire (RBQ; Moss & Oliver, 2008).

Demographic Questionnaire

The demographic questionnaire collected data on age, gender, diagnostic status, verbal ability and level of mobility.

Wessex Scale

Components of the Wessex Scale (Kuschlick, Blunden & Cox, 1973) were used to indicate degree of learning disability. The Wessex Scale is an informant based questionnaire that assesses daily living skills (feeding, dressing and washing), mobility, communication and literacy and has been used to assess adaptive behaviour in children and adults with learning disability. Percentage agreement on responses to each of these areas has demonstrated good inter-rater reliability and validity at subscale level for both children and adults (Kushlick et al., 1973; Palmer & Jenkins, 1979).

Challenging Behaviour Questionnaire (CBQ)

The CBQ (Hyman et al., 2002) is a brief informant based questionnaire regarding the presence or absence of self-injurious and physical aggression. The measure also examines eight topographies of self-injurious behaviour, adapted from Bodfish et al. (1995). Previous examination of the psychometric properties of the questionnaire has demonstrated good inter-rater reliability (Kappa value = .92) (Hyman et al., 2002).

Mood, Interest and Pleasure Questionnaire Short-form (MIPQ-S)

The MIPQ-S is a 12 item questionnaire, with scores on each item rated using a five point Likert scale. Items are scored by the participant's carer, based on observations during the previous two-week period. The MIPQ-S yields an overall score and scores on two-subscales, 'Mood' and 'Interest and Pleasure'. Low scores indicate low mood, interest and pleasure with a maximum overall score of 48. Examination of the psychometric of the MIPQ-S has demonstrated good test-retest and inter-rater reliability scores for the total and subscale scores, with kappa values for total

scores of .87 and .76 respectively. Internal consistency was .94. Evidence to support the construct validity was obtained by correlating scores with the Lethargy and Social Withdrawal scale on the Aberrant Behavior Checklist (Aman & Singh, 1986).

The Social Communication Questionnaire: Lifetime (SCQ-L)

The SCQ, developed by Rutter, Bailey and Lord, provides valuable information on a child's body movements, use of language or gestures, and style of interacting. The *Lifetime* version addresses a child's entire developmental history and comprises 40 items which assess the presence or absence of certain behaviours. There are three subscales: communication, social interaction and repetitive and stereotyped behaviours. Higher scores on these subscales indicate the presence of more abnormal behaviours. The questionnaire has been found to have good validity, closely matching the content of the Autism Diagnostic Interview and Observation Schedule (Berument, Rutter, Lord, Pickles, & Bailey, 1999; Howlin & Karpf, 2004).

The Activity Questionnaire (TAQ)

The TAQ (Burbidge & Oliver, 2008) is an informant questionnaire for use in relation to children and adults with a range of intellectual disability and is suitable for use with verbal and non-verbal, mobile and immobile individuals. Informants rate the frequency of behaviours that fall under the following headings: over activity, impulsivity and impulsive speech. Inter-rater reliability for verbal and non-verbal participants is .74 and .78 respectively with test-retest scores of .88 and .94 (verbal and non-verbal). Internal consistency is .94 (overall).

Repetitive Behaviour Questionnaire (RBQ)

The Repetitive Behaviour Questionnaire (RBQ; Moss & Oliver, 2008) is an informant questionnaire suitable for use with verbal and non-verbal children and adults with learning disabilities. The RBQ has five subscales: stereotyped behaviour; compulsive behaviour; insistence on sameness; restricted preferences and repetitive use of language. Informants rate the presence or absence of repetitive behaviour and for those behaviours that are present, informants then rate the frequency of occurrence, individual's response to interruption of the behaviour, level of interference that the behaviour has on the individual's everyday life and how much the individual insists on carrying out the behaviour. The RBQ has demonstrated good psychometric properties for individuals with severe and profound intellectual disabilities.

Procedure

A covering letter, information sheet, questionnaire pack, consent forms and a prepaid return envelope were sent to carers of prospective participants via the relevant syndrome support groups. Carers were asked to complete and return the consent form and questionnaire pack.

Data Analysis

Data distributions were examined and inspection of skew, kurtosis and results of Kolmogorov-Smirnov tests indicated that parametric tests could be used to examine age, MIPQ and TAQ scores. Scores on one subscale of the RBQ (Compulsive Behaviour) were not found to have a normal distribution for comparisons between individuals showing behaviours and individuals not showing behaviours within syndrome groups (Kolmogorov-Smirnov $Z = 2.35$; $p < .001$). Therefore, non-parametric tests were used to analyse this measure. On all other measures non-parametric tests were used due either to abnormal distribution or small sample size. The percentages of individuals showing self-injurious behaviour and physical aggression and different topographies of self-injury in the total sample and in each of the syndrome groups were derived from the CBQ.

Potential predictors of self-injurious behaviour were examined for each syndrome group separately. Within the 1p36 deletion syndrome group, participants showing self-injury or physical aggression were compared with those who did not show the behaviour. The Mann-Whitney test was used to examine the subscale scores of the MIPQ, TAQ and RBQ in relation to the presence of self injury and physical aggression.

Results

Demographic Characteristics

We compared the groups on key demographic characteristics and there were no significant differences for age, gender, self-help, verbal ability and mobility. However, comparisons between the groups using Chi-square tests for vision (χ (3) = 20.75, $p<.001$) and hearing (χ (3) = 17.23, $p<.001$) revealed significant differences between the groups. Post hoc contrasts showed significantly fewer individuals in the 1p36 deletion syndrome group with normal vision than individuals in the Angelman (χ (1) = 10.95, $p<.001$) and Cri du Chat syndrome groups (χ (1) = 12.24, $p<.001$). In addition a higher proportion of individuals in the Angelman syndrome group were found to have normal hearing when compared to individuals in the Cornelia de Lange syndrome group (χ (1) = 15.07, $p<.001$).

Prevalence of Self-Injury and Physical Aggression

Across the groups 54 (60.0%) showed self injury and 50 (55.6%) had showed physical aggression. Table 4 shows the number of participants showing self-injury and physical aggression in each of the four syndrome groups. Fourteen (60.9%) of the participants in the 1p36 deletion syndrome group showed self injury and twelve (52.2%) showed physical aggression. Chi-square analyses revealed no significant differences in prevalence between the four syndrome groups.

Table 4: Frequency and percentage of individuals showing self-injury and physical aggression in each group.

	Self-injurious behaviour	Physical aggression
	Frequency	Frequency
Syndrome Group	(%)	(%)
1p36	14	12
(n=23)	(60.9)	(52.2)
Angelman	8	18
(n=21)	(38.1)	(85.7)
Cri du Chat	16	16
(n=23)	(69.6)	(69.6)
Cornelia de Lange	16	4
(n=23)	(69.6)	(17.4)

Table 5 shows the number of participants showing different topographies of self-injury in each syndrome group. Chi-square analyses found that no specific topographies were more likely to be shown in any of the four syndrome groups. The most common topographies of self-injury reported in those with 1p36 deletion syndrome were 'bites self' (n=11, 47.8%) and 'hits self with body' (n=11, 34.8%).

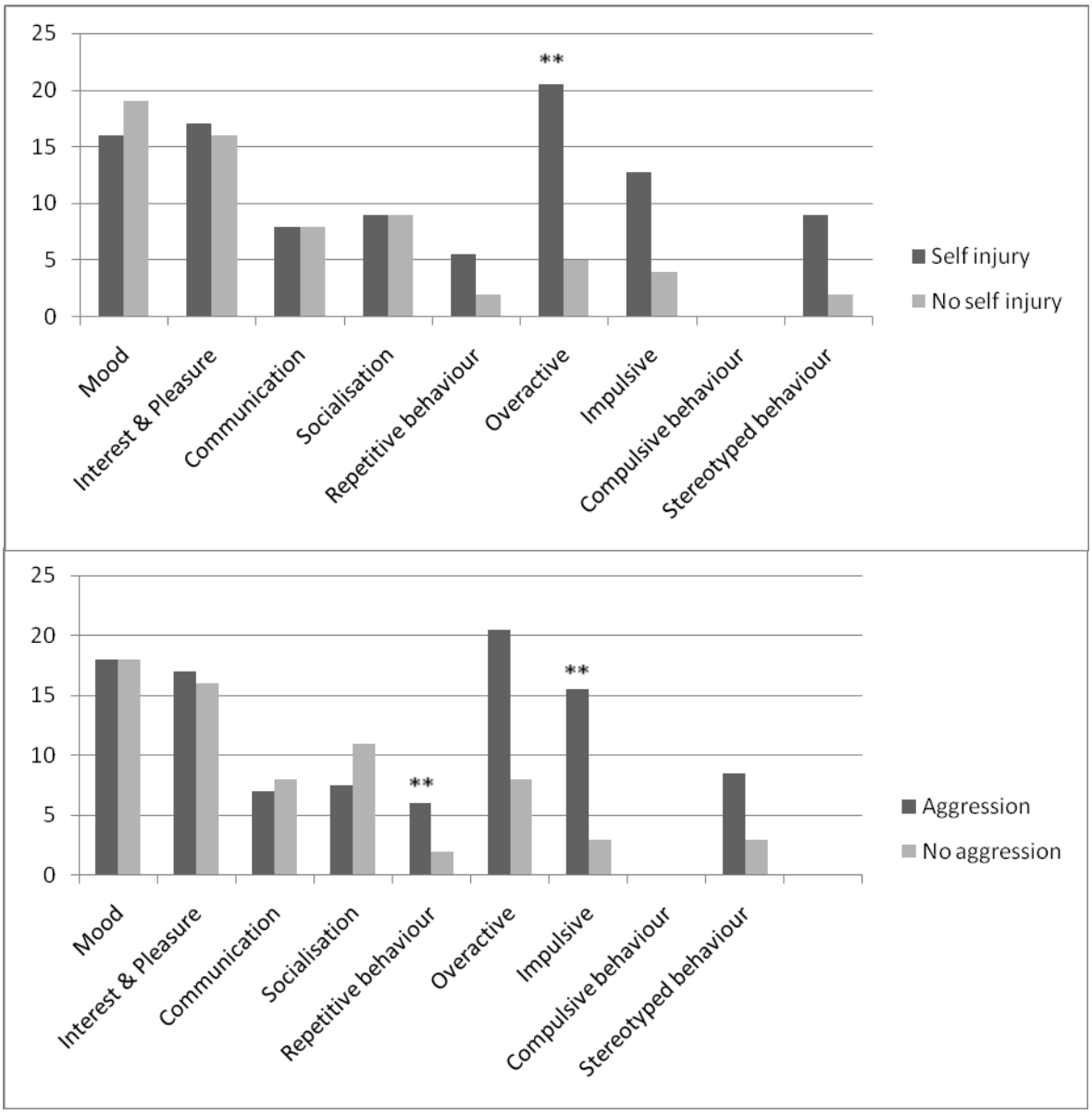
Table 5: Frequency and percentage of individuals showing different topographies of self-injury in each syndrome group.

	Topography Frequency (%)						
Syndrome Group	Hits self with body	Hits self against object	Hits self with object	Bites self	Pulls self	Rubs/ scratches self	Inserts
1p36 (n=23)	8 (34.8)	5 (21.7)	4 (17.4)	11 (47.8)	7 (30.4)	4 (17.4)	3 (13.0)
Angelman (n=21)	0 (0.0)	1 (4.8)	0 (0.0)	5 (23.8)	1 (4.8)	3 (14.3)	4 (19.0)
Cri du Chat (n=23)	7 (30.4)	9 (39.1)	4 (17.4)	5 (21.7)	10 (43.5)	5 (21.7)	3 (13.0)
Cornelia de Lange (n=23)	9 (31.9)	8 (34.8)	4 (17.4)	5 (21.7)	5 (21.7)	3 (13.0)	6 (26.1)

Predictors of Self-Injury and Physical Aggression within Groups

The second aim of the study was to examine differences in specific variables between individuals with and without self-injury and physical aggression within the 1p36 deletion syndrome group. Mann-Whitney tests were conducted in order to explore the differences in affect, autism spectrum behaviours, hyperactivity and repetitive behaviours between participants with and without self-injury and aggression. The results are shown in Figure, which shows that individuals with 1p36 with self-injury showed significantly higher scores on the Overactivity ($z = -3.22$; $p<.001$) and Stereotyped Behaviour ($z = -3.11$; $p<.001$) subscales than those who did not show SIB. Figure 1 also shows that individuals with 1p36 deletion syndrome with physical aggression were also found to score significantly higher on the Repetitive Behaviour ($z = -3.94$; $p<.001$) and Impulsivity subscales ($z = -3.19$; $p<.001$).

Figure 1: Bar charts to show differences in affect, autism spectrum behaviours, hyperactivity and repetitive behaviours in participants showing and not showing self-injurious (upper panel) and aggressive (lower panel) behaviour within the 1p36 syndrome group. (= significant at the p<.001 level)**



Summary

In summary**,** fourteen (60.9%) of the participants in the 1p36 deletion syndrome group showed self injury and twelve (52.2%) physical aggression. When compared to the other syndrome groups, no significant differences in prevalence of self-injurious or aggressive behaviour were found. The most common topographies of self-injury in the 1p36 deletion syndrome group were 'bites self' and 'hits self with body'. Again, when compared to the other syndrome groups, no significant differences in topographies of self-injury were found. Self-injury was associated with overactive and stereotyped behaviour. Aggressive behaviour was associated with high impulsivity and repetitive behaviour.

Study two

Aims

The second aim of the study was to employ experimental functional analysis with children with 1p36 deletion syndrome in order to examine operant influences, such as attention and demand, on self-injurious and aggressive behaviours. A further aim of the study was to examine the influence of social contact on affect via the occurrence of positive vocalisations (smiling, laughing, positive speech or facial expression) and negative vocalisations (crying, frowning, moaning, whining, screaming, negative speech or facial expression). The presence of proto-imperative communicative behaviours, in the form of approach and dissent, was also examined.

Method

Participants

Of the 23 individuals with 1p36 deletion syndrome included in study one, fourteen were reported to have displayed self-injurious or aggressive behaviour and so were contacted for further participation in the study. Of the fourteen individuals, six opted to participate. Of the eight participants who did *not* take part in the analogue observations, two opted out of the study with no reason given; one did not wish to participate as the participant had ceased self-injuring following completion of the questionnaires; two could not participate due to ill health, one lived out of the area covered by the research team (England), and two did not respond to written or telephone contact.

Of the six participants, two were male and four were female. Table 6 shows participant details for this sample for age and gender and standard scores for Communication, Daily Living Skills, Socialisation, Motor Skills and Adaptive Behavior Composite (calculated from the Vineland Adaptive Behavior Scale; Sparrow, Balla & Cicchetti, 2005).

Table 6: Demographics of the participants

Participant	Gender	Age[d]	Communication[e]	Daily Living Skills[e]	Socialisation[e]	Motor Skills[e*]	Adaptive Behaviour Composite[f]
1	Male	13yrs 11mths	40	47	45	-	43
2	Female	3yrs 3mths	47	46	63	31	44
3	Male	3yrs 2mths	79	64	72	59	65
4	Female	10yrs 11mths	48	50	57	-	50
5	Female	3yrs 3mths	59	62	63	54	55
6	Female	4 yrs 9mths	44	75	72	51	61

[d] Chronological age in years and months.

[e] Standard scores derived from the VABS.

[f] Mean age equivalent in months derived from the VABS

[*] For individuals aged 7.0 years and older the motor skills subdomain is not scored.

Setting

Participants were observed in a quiet room at their usual day placement (home, nursery or school). The aim was to carry out the observations in a familiar, informal and comfortable environment. On arrival, carers were briefed about the study and asked to complete the Vineland Adaptive Behavior Scale Parent/Caregiver Rating Form (Sparrow et al., 2005). Carers were invited to remain in a room next door throughout the observations.

Procedure

Throughout the experimental functional analysis two researchers were present, one to carry out the protocol and another to take a video record. The experimental functional analytic method used was based on that developed by Carr and Durand (1985) and has been used widely for ascertaining whether operant social reinforcement influences a given behaviour (e.g. Arron, Oliver, Hall, Sloneem, Forman & McClintock, 2006; Hall, Oliver & Murphy, 2001;Strachan et al. 2009). Each

of the six children was observed and videotaped whilst exposed to three conditions, each lasting 5 minutes: *High Attention* (A); *Low Attention* (B) and *Task Demand* (C). During the *High Attention* condition (A), which acted as a control condition, the researcher maintained a high level of both verbal and physical attention, such as talking, singing and clapping. The researcher remained in close proximity to the child (within arm's reach) and placed no demands upon the child. If the child displayed any self-injurious behaviour during condition A, the researcher ignored the behaviour and continued to maintain a high level of attention. In the *Low Attention* condition (B), the researcher maintained conversation with the second researcher and did not speak to or make eye contact with the child. The researcher began the condition in close proximity to the child and remained in this position even if the child moved away. If the child showed any self-injurious behaviour during condition B, the researcher gave brief (approximately 5-s) physical and verbal attention, requesting that the child stop the behaviour ("don't hurt yourself") and then continued to interact with the second researcher. During the *Task Demand* condition (C), the researcher requested the child to carry out a task that was challenging for them. The task was chosen based on parental report as well as information offered by the class teacher (if the observation was carried out in school). The child was initially given a verbal prompt to carry out the task, followed by the researcher modelling the task, and then giving a physical prompt (hand-over-hand guiding through the task). This process was completed until the condition ended. If the child displayed any self-injurious behaviour during condition C, the researcher gave verbal attention ("ok, you don't have to do this anymore"), then the researcher turned away from the child, removing the task. The researcher remained turned away from the child for a 10-s period, or until the child ceased the self-injurious behaviour. If the child showed further self-injurious behaviour the researcher turned away for a further 10-s period.

The *Low Attention* and *Task Demand* conditions were alternated between repeated presentations of the *High Attention* control condition. A series of conditions comprised eight *High Attention* (A) conditions, four *Low Attention* (B) and four *Task Demand* (C) conditions (ABACABACABACABAC). Obswin data collection software (Martin, Oliver, & Hall, 2001) was used to code the video recordings of each child and record the duration of a number of target behaviours.

Measurement and interobserver agreement

The behaviour of the researcher was recorded for 30% of the observations in order to evaluate the consistency of the independent variables. The percentage of time the researcher engaged in several behaviours was calculated for each condition: verbal attention (verbal contact from researcher), physical attention (physical contact initiated by researcher), verbal prompt (verbal demand to complete a task), model prompt (model of correct response by researcher) and physical prompt (hand over hand demand to complete a task). Total percentage of time for each condition could total over 100% as behaviours were not mutually exclusive. For condition A (high attention) perentage durations of the behaviours were : verbal attention 97.99; physical attention 27.23; verbal prompt 0; model prompt 0 and physical prompt 0. For condition B (low attention) verbal attention was 5.43; physical attention 4.11; verbal prompt 0; model prompt 0 and physical prompt 0. For condition C (task demand) verbal attention was 19.74; physical attention .75; verbal prompt 45.94; model prompt 15.66 and physical prompt 20.66. The data show that the integrity of the independent variables was maintained.

A second observer independently coded 25% of the observations. Interobserver agreement between the first and second observer were compared on a 10-s interval-by-interval basis, with agreements and disagreements scored on occurrence and non-occurrence for each behaviour category. Cohen's Kappa was employed in order to control for 'chance' levels of agreement. The Kappa coefficients for child codes were: approach (movement of participant to be within proximity of researcher, physical contact initiated by the participant) .80; physical dissent (moving away from the researcher, pulling away from physical contact or physical prompts) .62; positive vocalisations (smiling, laughing, positive speech or facial expression) .63; negative vocalisations (crying, frowning, moaning, whining, screaming, negative speech or facial expression) .74; aggression (aggressive behaviour e.g. hitting, kicking researcher or furniture) 1.00; destruct (destructive behaviour e.g. throwing) .73; head banging (banging head against chair, floor etc) .83; hand mouthing (participant's hand in own mouth) .80 and head hitting (hitting own head with own hand) .79. For researcher codes, the Kappa coefficients were: verbal attention .87; physical attention .83; verbal prompt .69; model prompt .61 and physical prompt .86. All indices were greater than .60 indicating that interobserver reliability was good (Landis & Kock, 1977).

Data Analysis

Effect size was calculated for each participant's self-injurious and aggressive behaviour across the experimental functional analysis conditions using a dominance statistic (*d,* Cliff; 1993), which yields an index of the extent to which one sample distribution differs from another. The *d* statistic

is a measure of how much one sample distribution lies above another and this is determined by comparing all scores in one condition to all scores in another using a dominance matrix. In order to calculate whether self-injury or aggression had an attention maintained function, each datum point (datum point = mean percentage time per trial) from the low attention condition was compared to every datum point from the high attention condition. Demand escape function was determined by comparing each task demand condition datum point to every datum point from the high attention conditions.

An example of a calculated dominance matrix is shown in Table 7. The cells in Table 7 are calculated by endorsing a value of +1 if the row value (low attention datum point) is greater than the column value (high attention datum point), -1 if the row value is less than the column value and zero if the two values are identical. The sample value *d* is the mean value of the elements in the dominance matrix. A value of +1 would indicate that every datum point in a series is greater than every other datum point in another series. A value of -1 would indicate that every datum point in a series is less than every other datum point in another series.

Table 7: Example of a dominance matrix from participant 2 for head hitting.

	High attention							
Low attention	3.95	0	0	2.99	2.31	0.66	6.64	1
4.61	1	1	1	1	1	1	1	-1
5.32	1	1	1	1	1	1	1	-1
3.65	-1	1	1	1	1	1	1	-1
1.32	-1	1	1	-1	-1	1	1	-1

Calculation: 16/32= .50

d= .50 Moderate attention maintained function.

In order to compare the *d* statistic for social function at a categorical level, a cut-off value was arbitrarily nominated. A *d* value of between 0 and .33 indicates no social function, whilst a *d* value of above .33 indicates a moderate function for a behaviour and a *d* value above .66 indicates a strong function for a behaviour. It has been demonstrated that this statistic is robust when employed with non-normal distributions, even when variances differ (Cliff, 1993).

Results

In order to describe the overall levels of behaviour in each participant, the total percentage of time that participants engaged in each behaviour was aggregated across conditions. These percentages are shown in Table 8. Table 8 shows that hand mouthing was the most common topography of self-injury. In two of the participants self-injurious behaviour was not observed across analogue conditions even though it had been previously reported in the questionnaire study.

Behaviour % Duration									
Participant	Approach	Dissent	Positive vocalisations	Negative vocalisations	Non-contact stereotypy	Hand mouth	Hand head	Bang head	Aggression
1	3.40	.56	1.62	1.99	-	32.98	-	.73	.17
2	7.83	2.92	.90	.47	-	23.04	2.63	-	-
3	1.32	.47	9.79	27.66	.04	16.80	-	-	-
4	.61	.44	23.25	12.99	55.25	1.96	-	-	-
5	8.44	3.26	6.79	14.67	2.21	-	-	0	.42
6	1.95	1.41	8.01	.17	-	-	-	0	-

Table 8: Total percentage duration of behaviours across all conditions for participants (- indicates where a behaviour was not reported).

Self injury and aggression

To examine the function of self-injurious behaviour, individual plots, shown in Figure 2, were produced. The data in Figure 2 show that three of the five participants who showed self-injury or aggression showed the behaviour for more than 10% of the time during any one condition. Participants 1, 2, 3 and 4 showed self-injurious behaviours across all three conditions.

Cliff's *d* statistics for attention and demand were calculated for each behaviour for each participant. For hand mouthing, participant 1 showed a strong attention maintained function and participant 4 a moderate attention maintained function. Participant 2 showed a moderate attention maintained function for head hitting. For participant 2, hand mouthing was significantly *more* likely to occur in the *High Attention* condition than in the *Low Attention* condition. For participant 1, head banging was moderately *less* likely to occur in the *Task Demand* condition compared with the *High Attention* condition. For participant 3, hand mouthing was significantly *more* likely to occur in the *High Attention* condition than in both the *Low Attention* and *Task Demand* conditions. Participant 1 showed a moderate effect for aggression in the *High Attention* condition and this is not consistent with a conventional functional interpretation.

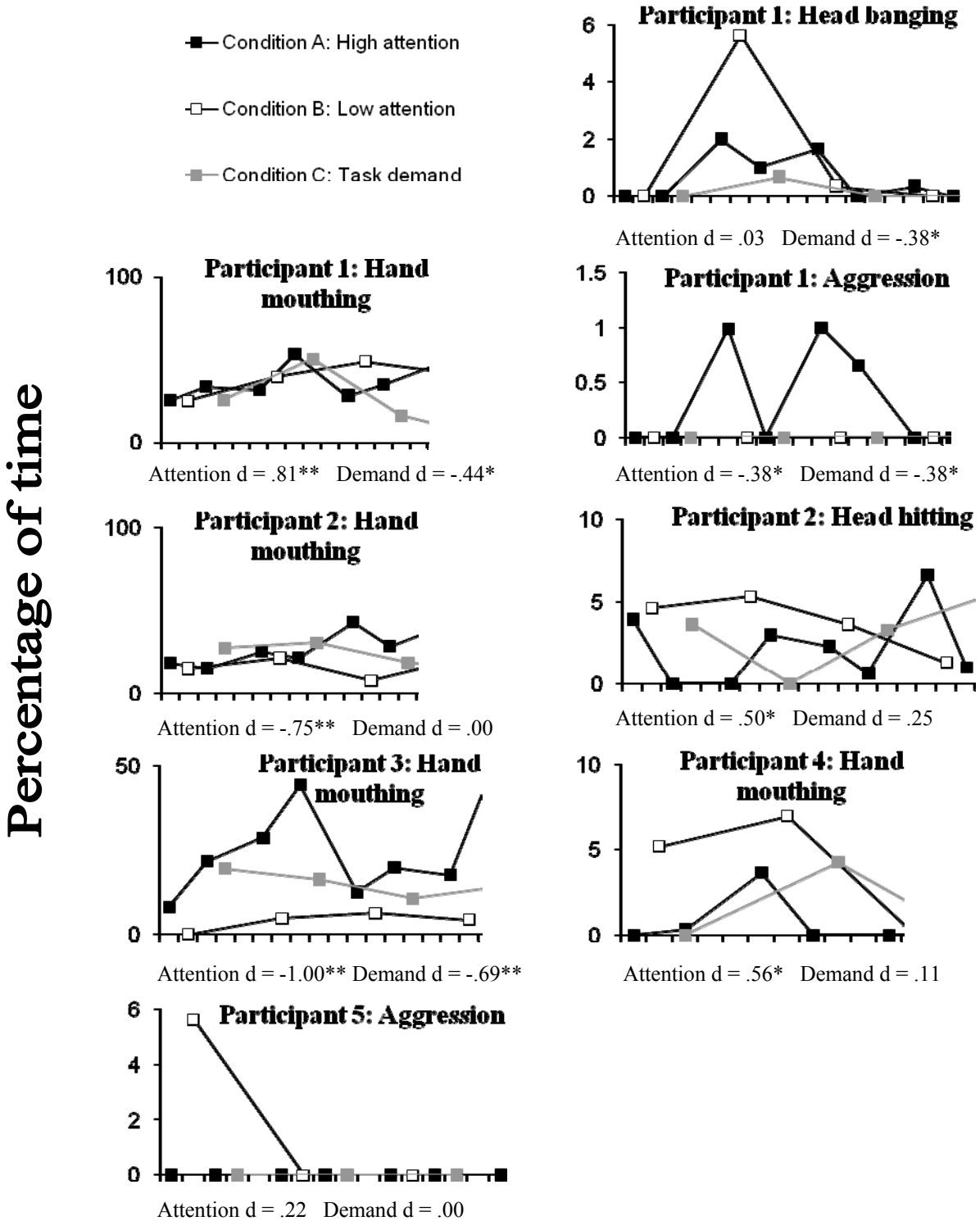


Figure 2: Percentage of time participants engaged in self-injurious/aggressive behaviour across conditions. Cliff's *d* statistics for Attention and Demand are shown below each graph (* = moderate function; ** = strong function).

Positive and Negative Vocalisations

To explore affect via the occurrence of positive and negative vocalisations, individual plots were created to show the percentage of time each child spent engaged in these behaviours for each condition. These plots are shown in Figure 3. All six participants showed higher levels of positive vocalisations during the *High Attention* conditions compared with the *Low Attention* and *Task Demand conditions*. However, it must be noted that participants 1 and 2 engaged in either positive or negative vocalisations for less than 10% of the time across all conditions. Participants 3, 4 and 6 showed relatively high levels of positive vocalisations. Participants 1, 2, 3 and 5 showed a higher level of negative vocalisations in the *Low Attention* condition compared with the *High attention* and *Task Demand* conditions. Participant 4 showed a higher level of negative vocalisations in the *Task Demand* condition when compared with the *High* and *Low Attention* conditions.

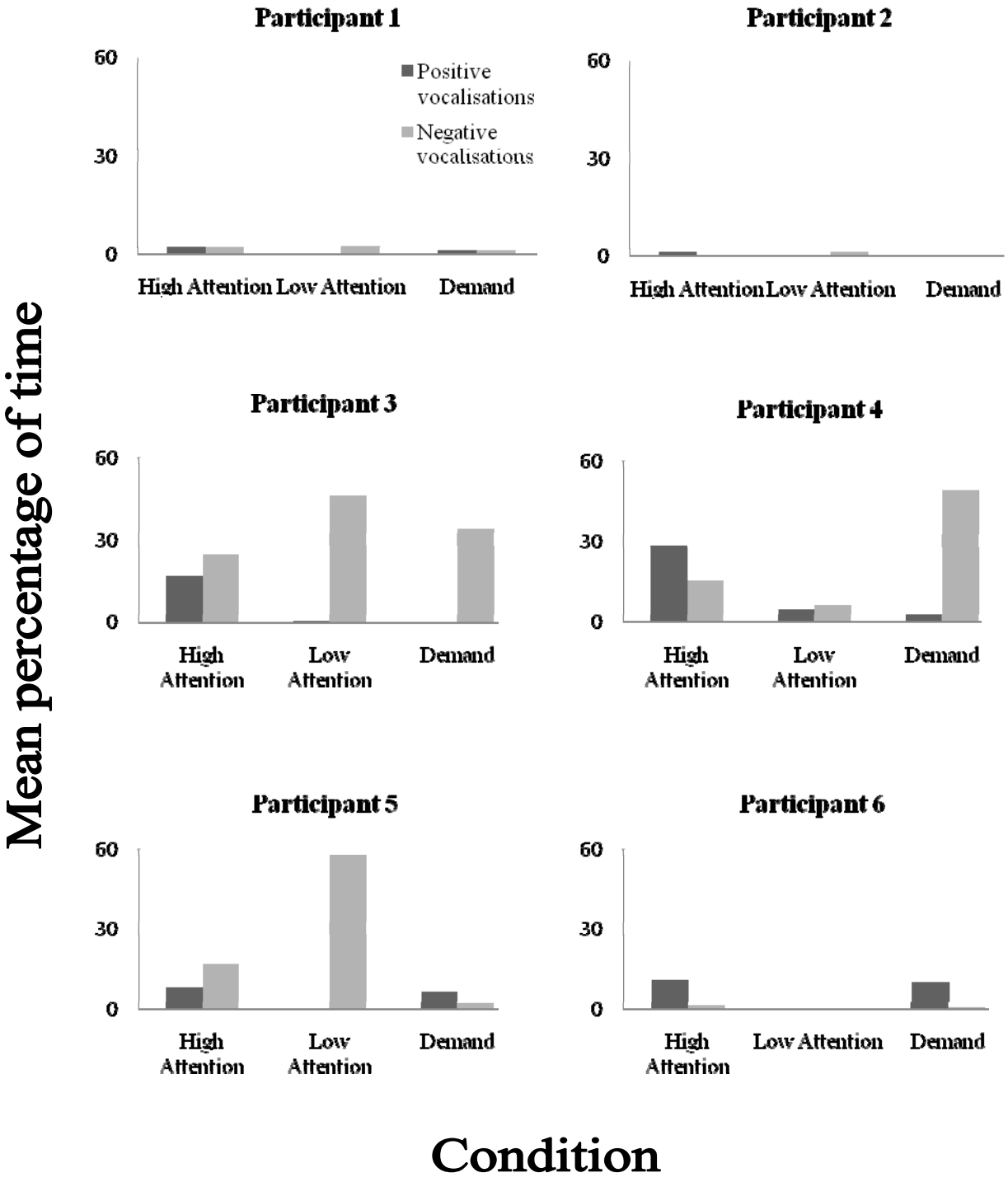


Figure 3: Percentage of time participants showed positive and negative vocalisations across conditions.

Proto imperative communicative behaviours

The occurrence of proto imperative communicative behaviours in the form of approach and dissent was examined across each of the conditions. Individual plots were created to show the percentage of time each child spent engaged in these behaviours across the three conditions. These plots are shown in Figure 4. All of the participants showed some level of approach and dissent behaviours. Five of the six participants displayed higher percentages of approach behaviours compared to dissent behaviours in the *High Attention* condition. All six participants elicited approach behaviours during the *Low Attention* condition. Participants 4 and 5 showed the highest levels of dissent behaviours in the *Task Demand* conditions, whereas participants 2, 3 and 5 showed higher levels of dissent behaviours in the *High Attention* condition.

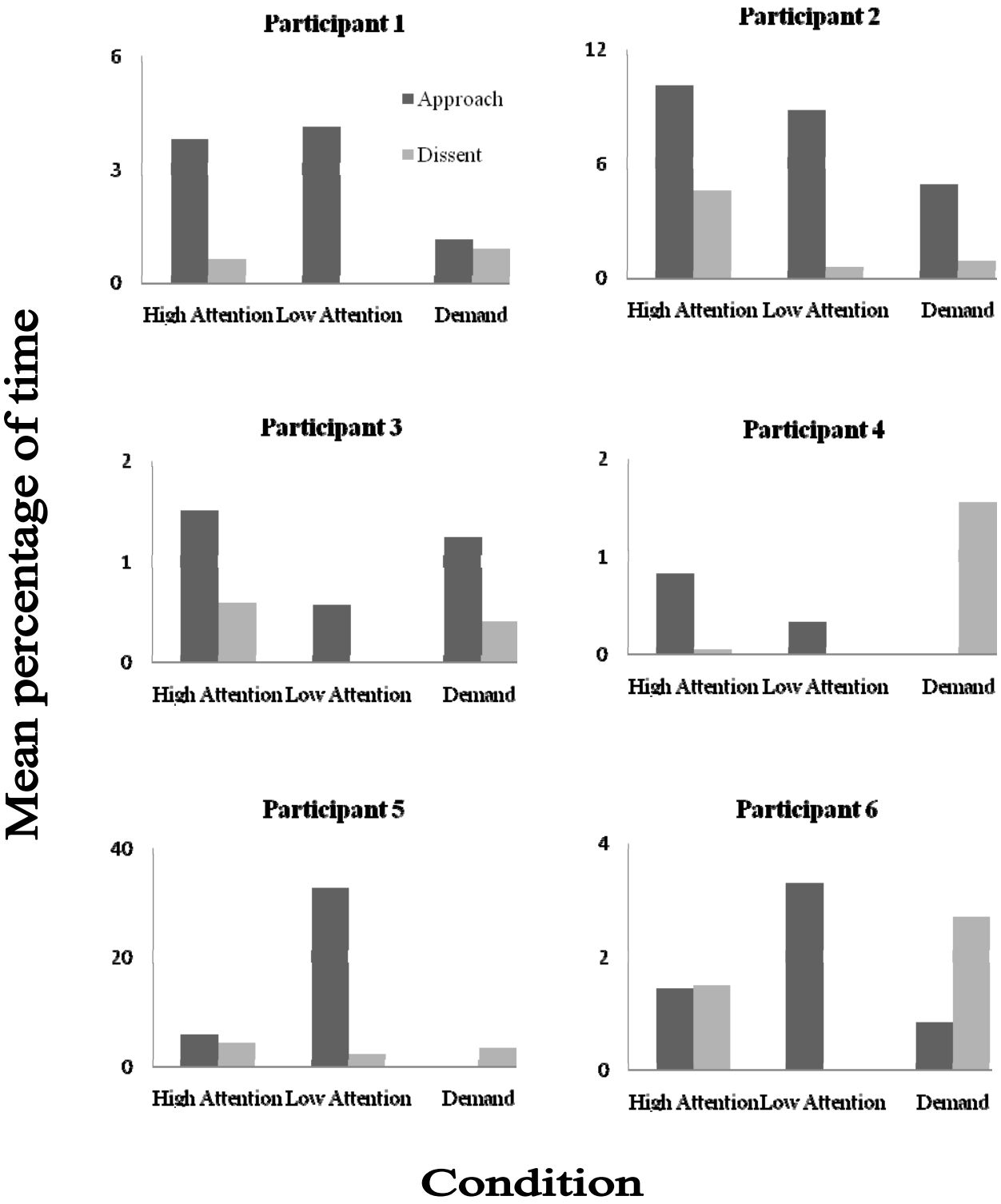


Figure 4: Percentage of time participants engaged in approach and dissent behaviour across conditions.

Summary

In summary, high levels of hand mouthing were shown by two participants. Two participants showed no self injury although the behaviour was reported in the survey stage. Two participants showed aggressive behaviours, one of these in a high risk condition (low attention). For three participants there was evidence of attention maintained function, two for hand mouthing, and one for head hitting. For no participants was there a demand escape function. Three participants showed relatively high levels of positive vocalisations (smiling, laughing, positive speech or facial expression). All participants showed some level of proto-imperative approach and dissent behaviours with five of the six participants displaying higher levels of approach behaviours compared to dissent behaviours in the *High Attention* condition. All six participants evidenced approach behaviours during the *Low Attention* condition.

Discussion

Principal findings

This was the first study to explore the prevalence and aetiology of self-injurious and aggressive behaviours in individuals with 1p36 deletion syndrome. Reliable, valid questionnaire measures were used to examine the prevalence of self-injury and aggression in individuals with 1p36 deletion syndrome alongside matched comparison groups. Carer reports indicated that 14 (60.9%) of the participants in the 1p36 deletion syndrome group showed self injury and 12 (52.2%) showed physical aggression, with self biting found to be the most common topography of self-injury. Self-injurious behaviour was found to be associated with overactivity and stereotyped behaviour and aggression was associated with impulsivity and repetitive behaviour. Experimental functional analysis was employed to examine potential environmental determinants of self injury and aggression. This was a robust experimental method and interobserver reliability was good. Behavioural data confirmed high levels of hand mouthing in two participants and showed that for three participants there was evidence of attention maintained function of self injury. Examination of the influence of social contact on affect found that five of the six participants displayed higher levels of positive vocalisations compared to negative vocalisations in the *High Attention* condition. Three participants showed relatively high levels of positive vocalisations. Exploration of proto-imperative communicative behaviours found all participants to show some level of approach and dissent behaviour. Five of the six

participants displayed higher levels of approach behaviours compared to dissent behaviours in the *High Attention* condition. All six participants evidenced approach behaviours during the *Low Attention* condition.

Comparison with previous literature

The high prevalence of self injury and aggression in the 1p36 deletion syndrome group is consistent with previous research findings indicating that self injurious behaviour, is a common feature in individuals with 1p36 deletion syndrome (Battaglia, 2005). However, the small sample size should be noted. The most common topography of self-injury in the 1p36 deletion syndrome group was self-biting and this is consistent with previous research findings that sucking fingers excessively, hand/wrist biting and hand chewing are commonly seen in individuals with 1p36 deletion syndrome (Battaglia, 2005). Hand mouthing has also been found to be common among other syndrome groups, such as Lesch-Nyhan, Rett, Cornelia de Lange and Fragile X syndromes (Anderson & Ernst, 1994; Hall, Oliver & Murphy, 2001; Hyman et al., 2002; Symons, Clark, Hatton, Skinner & Bailey, 2003; Arron, et al., in press).

The overall prevalence of self-injury in some syndrome groups in general has been found to be high, with existing literature indicating that Fragile X, Prader Willi and Smith Magenis syndromes are all associated with self-injury (Clarke et al., 1996; Dykens and Smith 1998; Finucane, Dirrigl and Simons, 2001; Symons et al., 2003). In participants with 1p36 deletion syndrome self-injurious behaviour was associated with overactivity and stereotyped behaviour. This finding is consistent with previous research that has found overactivity and stereotyped behaviour to be predictors of self-injury among individuals with intellectual disabilities and some genetic syndromes (Arron, et al., in press; Bradley, Summers, Wood and Bryson, 2004; Marston, Perry and Roy, 1997; Oliver et al. 2008; Petty & Oliver, 2005; Rojahn, Matson, Naglieri and Mayville, 2004). Aggressive behaviour in the 1p36 deletion syndrome group was found to be associated with impulsivity and repetitive behaviour. Again, this finding is consistent with existing literature demonstrating impulsive and repetitive behaviours as predictors of aggression (Arron, et al., 2009; Clarke et al. 2002; Hyman et al., 2002).

Whilst a high prevalence of hand mouthing was found, and thus appears to be part of a behavioural phenotype, the environment was found to have an influence for two participants, where there was evidence of attention maintained function. This finding is consistent with

previous research which has demonstrated an environmental effect on self-injury in individuals with genetic syndromes such as Lesch-Nyhan, Cornelia de Lange, Smith-Magenis and Rett syndromes (Arron, Oliver, Hall, Sloneem, Forman & McClintock, 2006; Hall, Oliver & Murphy, 2001; Moss et al., 2005; Taylor and Oliver, 2008). It also provides further evidence for placing emphasis on environmental determinants of self injurious-behaviour.

Three participants showed an effect in the *High Attention* condition which as previously noted is not consistent with a conventional functional interpretation. Participants 2 and 3 both showed a strong effect for hand mouthing and participant 1 showed a moderate effect for aggression. This means that more of these behaviours were displayed in the *High Attention* condition compared with the *Low Attention* condition. This can be interpreted in two ways. Higher rates of challenging behaviour occurring in periods of high attention may suggest that the behaviour serves to escape from social interaction with others, a form of 'social escape'. Alternatively, it is possible that the behaviour occurs more frequently as a way of *increasing* social attention from others. Both of these patterns suggest that participants may be making some strong associations between their behaviour and the surrounding environment which in turn suggests that the way in which other people respond to the self-injurious or aggressive behaviour is very important. Although this remains inconclusive, the significant difference between hand mouthing in the *High Attention* condition compared with the *Low Attention* and *Task Demand* conditions again demonstrates an environmental influence on the behaviour.

Anecdotal reports of high sociability in individuals with 1p36 deletion syndrome prompted exploration of the influence of social contact on affect and social approach. Three participants showed relatively high levels of positive vocalisations and all participants showed higher levels of positive vocalisations in the high attention condition compared with the low attention and task demand conditions. Participant 4 in particular showed very high levels of smiling and laughing. Horsler and Oliver (2006b) reviewed 64 studies on 842 cases of Angelman syndrome and found that 56 studies (88%) made reference to high levels of laughing, smiling or happy demeanour in participants. Recent experimental research has also demonstrated that in children with Angelman syndromes smiling and laughing are influenced by the level and type of social interaction (Horsler & Oliver, 2006b; Oliver, Demetriades & Hall, 2002; Strachan, Shaw, Burrow, Horsler, Allen & Oliver, 2009;). For example, in comparison to a contrast group, children with Angelman syndrome showed a greater increase in laughing and smiling in response to social interaction and actively sought social interaction

with adults (Oliver, Horsler, Berg, Bellamy, Dick, & Griffiths, 2007). Similarly, it may be that children with 1p36 deletion syndrome have a tendency to seek out social interaction, suggesting that they find interaction pleasurable.

Study strengths and limitations

The prevalence data should be considered in relation to the method used to recruit participants in this study. Participants were recruited via syndrome support groups and it has been hypothesised that families caring for individuals showing challenging behaviours are more likely to join support groups (Hyman et al., 2002). However, if this is the case, then this bias is comparable across groups and therefore comparisons of self-injurious and aggressive behaviour between the syndrome groups remains valid. As 1p36 deletion syndrome is rare, consequently the number of participants was small. However, the study provides the first exploration of self-injury and aggression in 1p36 deletion syndrome using behavioural assessment methods of known psychometric properties and experimental functional analysis. The procedure would have been more robust had further repetitions of each condition been carried out, however, it was felt that this may place unnecessary stress on the participants. Research into the effects of familiar versus unfamiliar adults when carrying out experimental functional analysis with children with intellectual disabilities has suggested that higher rates of problem behaviours have been produced when using the child's caregiver in comparison to an unfamiliar researcher (English & Anderson, 2004). Indeed, anecdotal reports from carers in this study indicated that participants tended to show higher level of self-injury and aggression when at they were at home with familiar caregivers than when they were in a less familiar environment with unfamiliar adults. Future research in this area may benefit from this comparison in order to gain more accurate data on the prevalence and causes of self-injury in this group.

Clinical implications and future directions

Self-injurious behaviour can impact significantly on the quality of life of an individual and their family and successful intervention remains an ongoing challenge to professionals. It is therefore of importance that a greater understanding of the predictors and risk markers for self-injury is gained by both caregivers and professionals working with this population. This may then act to increase awareness of potential problems that might be faced and assist with provision of early intervention where necessary.

Children with 1p36 deletion syndrome have been found to have a poor communicative repertoire, with less than 40% of the children in this study showing verbal/signing ability (Marr, 2009). It has been suggested that certain behaviours shown by individuals with intellectual disability, for example stereotyped behaviours, are not only associated with, but are also precursors of self-injury (Symons , Sperry, Dropik & Bodfish, 2005). This finding is consistent with the view that self injury results from an interaction between biological and environmental determinants. It also demonstrates the importance of shaping precursor behaviours to have a communicative function before they result in self-injury. Carrying out methodologically robust research will help to further clarify predictive risk markers in order to inform the phenomenology, assessment and treatment of self-injurious behaviour in this population.

References

Aman, M.G., & Singh, N. N. (1986). *Manual for Aberrant Behavior Checklist*. East Aurora, NY: Slosson Educational Publications.

Anderson, L. T., & Ernst, M. (1994). Self-injury in Lesch-Nyhan disease. *Journal of Autism and Developmental Disorders, 24,* 67-81.

Arron, K., Oliver, C., Hall, S., Sloneem, J., Forman, D., & McClintock, K. (2006). Effects of Social Interaction on Pragmatic Communication and Self-Injurious Behavior in Cornelia de Lange syndrome. *American Journal on Mental* Retardation*, 111*, 184-192.

Arron, K., Oliver, C., Berg, K., Moss, J., & Burbidge, C. (in press). Delineation of behavioural phenotypes in genetic syndromes: 2. Prevalence, phenomenology and correlates of self-injurious and aggressive behaviour. *Journal of Autism and Developmental Disorders.*

Battaglia, A. (2005). Deletion 1p36 syndrome: a newly emerging clinical entity. *Brain and Development*, 27, 358-361.

Berument, S. K., Rutter, M., Lord, C., Pickles, A., & Bailey, A. (1999). Autism Screening Questionnaire: Diagnostic validity. *British Journal of Psychiatry, 175*, 444-451.

Bodfish, J. W., Crawford, T. W., Powell, S. B., Parker, D. E., Golden, R. N., & Lewis, M. H. (1995). Compulsions in Adults with Mental-Retardation - Prevalence, Phenomenology and Comorbidity with Stereotypy and Self-Injury. *American Journal on Mental Retardation, 100*, 183-192.

Bradley, E., Summers, J., Wood, H., & Bryson. (2004). Comparing rates of psychiatric and behavior disorders in adolescents and young adults with severe intellectual disability with and without autism. *Journal of Autism and Developmental Disorders, 34(2*), 151-161.

Burbidge, C., & Oliver, C. (2008). Activity Questionnaire: *Manual for administration and scorer interpretation.* University of Birmingham.

Chapman, R.S., & Hesketh, L. (2000). The behavioral phenotype of Down syndrome. *Mental Retardation and Developmental Disabilities Research Review*, *6*, 84-95.

Clarke, D. J., Boer, H., Chung, M. C., Sturmey, P., & Webb, T. (1996). Maladaptive behaviour in Prader-Willi syndrome in adult life. *Journal of Intellectual Disability Research 40* (2)*,* 159–165.

Clarke, D. J., Boer, H., Whittington, J., Holland, A., Butler, J., & Webb, T. (2002). Prader-Willi syndrome, compulsive and ritualistic behaviours: the first population-based survey. *British Journal of Psychiatry*, *180*, 358-362

Cliff, N. (1993). Dominance statistics: ordinal analyses to answer ordinal questions. *Psychological Bulletin, 114,* 494-509.

Collins, M. S. R., & Cornish, K. (2002). A survey of the prevalence of stereotypy, self-injury and aggression in children and young adults with Cri du Chat syndrome. *Journal of Intellectual Disability Research, 46* (2), 133–140.

Cooper, S. A., Smiley, E., Allan, L. M., Jackson, A., Finlayson, J., Mantry, D., & Morrison, J. (2009). Adults with intellectual disabilities: prevalence, incidence and remission of self-injurious behaviour, and related factors. *Journal of Intellectual Disability Research*, *53*(3), 200-16.

Cook, F. (2009). An investigation of sociability: Delineating a behavioural and social phenotype for Monosomy 1p36 Deletion Syndrome. Unpublished D. Clin. Psych thesis. University of Birmingham, Birmingham, England.

Deb, S., Thomas, M., & Bright, C. (2001). Mental disorder in adults with intellectual disability: Prevalence of functional psychiatric illness among a community-based population aged between 16 and 64 years. *Journal of Intellectual Disability Research, 45*, 495–505.

Dykens, E. M., & Kasari, C. (1997). Maladaptive behavior in children with PWS, Down's syndrome, and nonspecific mental retardation. *American Journal on Mental Retardation*, *102* (3), 228-237.

Dykens, E. M., & Smith, A. C. M. (1998). Distinctiveness and correlates of maladaptive behaviour in children and adolescents with Smith-Magenis syndrome. *Journal of Intellectual Disability Research*, 42, 481-489.

English, C. L. & Anderson, C. M. (2004). Effects of familiar versus unfamiliar therapists on responding in the analogue functional analysis. *Research in Developmental Disabilities, 25,* 39-55.

Finucane, B., Dirrigl, K. H., & Simon, E. W. (2001). Characterization of self-injurious behaviors in children and adults with Smith-Magenis syndrome. *American Journal on Mental Retardation, 106*, 52-58.

Green, L., & Freed, D. E. (1993). The substitutability of reinforcers. *Journal of the Experimental Analysis of Behavior, 60,* 141-158.

Hall, S. S., Oliver, C., & Murphy, G. (2001). Self-injurious behaviour in young children with Lesch-Nyhan Syndrome. *Developmental Medicine and Child Neurology, 43* (11), 745-749.

Hanley, G. P., Iwata, B. A., & McCord, B. E. (2003). Functional analysis of problem behaviour: A review. *Journal of Applied Behavior Analysis, 36*, 147-185.

Heilstedt, H.A., Ballif, B. C., Howard, L. A., Lewis, R. A., Stal, S., Kashork, C. D., Bacino, C. A., Shapira, S. K., & Shaffer, L. G. (2003) Physical Map of 1p36, Placement of Breakpoints in Monosomy 1p36, and Clinical Characterization of the Syndrome. *American Journal of Medical Genetics, 70*, 409-412.

Horsler, K. J. & Oliver, C. (2006). The behavioural phenotype of Angelman syndrome, *Journal of Intellectual Disability Research, 50*, 33-53.

Horsler, K. J., & Oliver, C. (2006). Environmental influences on the behavioural phenotype of Angelman syndrome. *American Journal on Mental Retardation, 11*, 311-321.

Hyman, P., Oliver, C., & Hall, S. (2002). Self-injurious behavior, self-restraint and compulsive behaviors in Cornelia de Lange syndrome. *American Journal on Mental Retardation, 107,* 146-154.

Iwata, B. A., Dorsey, M. F., Slifer, K. J., Bauman, K.E., & Richman, G. S. (1982). Toward a functional analysis of self-injury. *Journal of Applied Behaviour Analysis, 27,* 197-209.

Knight-Jones E., Knight S.J.L., Heussler H., Kyne L., Regan R., Flint J., & Martin K. (2000). Neurodevelopmental profile of a new dysmorphic syndrome associated with submicroscopic partial deletion of 1p36.3. *Dev Med Child Neurol.* 42. 201-206

Kushlick, A., Blunden, R., & Cox, G. (1973). A method of rating behaviour characteristics for use in large scale surveys of mental handicap. *Psychological Medicine, 3*, 466-78.

Lovaas, O. I., Freitag, G., Gold, V. J., & Kassorla, I. C. (1965). Experimental Studies in Childhood Schizophrenia: Analysis of Self-Destructive Behavior. *Journal of Experimental Child Psychology, 2,* 67-84.

Marr, A. (2009). Delineation of the behavioural phenotype of 1p36 deletion syndrome: Prevalence, correlates and experimental functional analysis of self-injurious and aggressive behaviour. Unpublished D. Clin. Psych thesis. University of Birmingham, Birmingham, England.

Marston, G. M., Perry, D. W., & Roy, A. (1997). Manifestations of depression in people with intellectual disability. *Journal of Intellectual Disability Research*, *41*, 476-480.

McClintock, K., Hall, S., & Oliver, C. (2003). Risk markers associated with challenging behaviours in people with intellectual disabilities: a meta-analytic study. *Journal of Intellectual Disability Research, 47,* 405-416.

Moss, J., Oliver, C., Hall, S., Arron, K., Sloneem, J., & Petty, J. (2005*)* The association between environmental events and self-injurious behaviour in Cornelia de Lange syndrome. *Journal of Intellectual Disability Research, 40*, 69-77.

Moss, J., Oliver, C., Arron, K., Burbidge, C., & Berg, K. (2009). The prevalence and phenomenology of repetitive behaviour in genetic syndromes. *Journal of Autism and Developmental Disorders, 39* (4), 572-88.

Moss, J., & Oliver, C. (2008). Repetitive Behaviour Questionnaire: *Manual for administration and scorer interpretation.* University of Birmingham.

Oliver, C. (1995). Self-injurious behaviour in children with learning disabilities: Recent advances in assessment and intervention. *Journal of Child Psychology and Psychiatry, 30*, 909-927.

Oliver, C., Demetriades, L., & Hall, S. (2002). The effect of environmental events on smiling and laughing behavior in Angelman Syndrome. *American Journal on Mental Retardation, 107* (3), 194-200.

Oliver, C., Hall, S., & Murphy, G. (2005). The early development of self-injurious behaviour: Evaluating the role of social reinforcement. *Journal of Intellectual Disability Research*, 49, 591-599.

Oliver, C., Arron, K., Hall, S., & Sloneem, J. (2008). The behavioral phenotype of Cornelia de Lange syndrome: case control study. *The British Journal of Psychiatry, 193,* 466-470.

Kau, A. S. M., Reider, E. E., Payne, L., Meyer, W. A., & Freund, L. (2000) Early behaviour signs of psychiatric phenotypes in fragile X syndrome. *American Journal on Mental Retardation, 105* (4), 286-299.

Palmer, J., & Jenkins, J. (1982). The 'Wessex' behaviour rating system for mentally handicapped people: Reliability study. *British Journal of Mental Subnormality, 28*, 88–96.

Petty, J., & Oliver, C. (2005). Self-injurious behaviour in people with intellectual disability. *Current Opinion in Psychiatry, 18*, 484-489.

Powell, S. B., Bodfish, J. W., Parker, D., Crawford, T. W., & Lewis, M. H. (1996). Self-restraint and self-injury: Occurrence and motivational significance. *American Journal on Mental Retardation, 101*, 41-48.

Reish, O. K., Berry, S., & Hirsch, B. (1995) Partial monosomy of chromosome. 1p36.3. *American Journal of Medical Genetics, 59*, 467–75.

Rojahn, J., Matson, J. L., Naglieri, J. A., & Mayville, E. (2004). Relationships between psychiatric conditions and behavior problems among adults with mental retardation. *American Journal on Mental Retardation*, *109*, 21-33.

Ross, E., & Oliver, C. (2002). The relationship between levels of mood, interest and pleasure and "challenging" behaviour in adults with severe and profound intellectual disabilities. *Journal of Intellectual Disability Research, 46*(3), 191-197.

Rutter, M., Bailey, A., & Lord, C. (2003). *SCQ: The Social Communication Questionnaire*. Manual. Western Psychological Services: Los Angeles, CA.

Shaffer, L.G., & Lupski, J.R. (2000). Molecular mechanisms for constitutional chromosomal rearrangements in humans. *Annual Review of Genetics*, 34, 297-329

Shapira, S.K., McCaskill, C., Northrup, H., Spikes, A.S., Elder, F.F.B., Reid Sutton, V., Korenberg, J.R., Greenberg, F., & Shaffer, L.G. (1997). Chromosome 1p36 deletions: The clinical phenotype and molecular characterization of a common newly delineated syndrome. *American Journal of Human Genetics*. *61*, 642-650

Slavotinek, A., Shaffer, L. G., & Shapira, S.K. (1999). Monosomy 1p36. *Journal of Medical Genetics*, *36*, 657-663.

Sparrow, S.S., Cicchetti, D.V., & Balla, D.A., (2005). *Vineland Adaptive Behavior Scales: Second Edition (Vineland II), Survey Interview Form/Caregiver Rating Form*, Livonia, MN: Pearson Assessments.

Strachan, R. S., Shaw, R., Burrow, C., Horsler, Allen, D., & Oliver, C. (2009). Experimental functional analysis of aggression in children with Angelman syndrome. *Research in Developmental Disabilities, 30* (5), 1095-1106.

Symons, F. J., Clark, R. D., Hatton, D. D., Skinner, M., & Bailey, D. B. (2003). Self-injurious behavior in young boys with fragile X syndrome. *American Journal of Medical Genetics Part A*, *118a*, 115-121.

Symons, F. J., Sperry, L. A., Dropik, P. L., & Bodfish, J. W. **(**2005). The early development of stereotypy and self-injury: A review of research methods. *Journal of Intellectual Disability Research, 49* (2), 144-58.

Taylor, L., & Oliver, C. (2008). The behavioural phenotype of Smith-Magenis syndrome: Evidence for a gene-environment interaction. *Journal of Intellectual Disability Research, 52*, 830-841.

Udwin, O. & Yule, W. (1991). A cognitive and behavioral phenotype in Williams syndrome. *Journal of Clinical and Experimental Neuropsychology*, *13*, 232-244.

Woodcock, K., Oliver, C. & Humphreys, G.W. (2009). Associations between repetitive questioning, resistance to change and temper outbursts and anxiety in Prader-Willi and Fragile-X syndromes. *Journal of Intellectual Disability Research, 53* (3), 265-278.

Executive Summary: Self-injury in 1p36 deletion syndrome

What is 1p36 deletion syndrome?

1p36 deletion syndrome is a chromosome disorder, which means that it is caused by a missing piece of genetic material, or DNA. We have 46 chromosomes and 1p36 deletion syndrome is caused by a missing part of a chromosome called band 1p36. Between 1 in 5,000 and 1 in 10,000 babies are born with 1p36 deletion syndrome with twice the number of girls compared to boys. Children with 1p36 deletion syndrome have a lot of physical and medical problems such as growth and feeding difficulties, seizures, vision and hearing problems and heart problems. They are also delayed in their development which results in a number of problems including difficulties walking, communicating and learning.

Some researchers have looked at the physical problems that people with 1p36 deletion syndrome suffer from, but far less attention has been paid to the behavioural problems. We know that people with 1p36 deletion syndrome show some challenging behaviours, such as aggression and self-injury and we wanted to find out more about this. Parents of children with 1p36 deletion syndrome also report that their children are very affectionate and sociable and we wanted to find out more about this as well.

Why did we decide to carry out this study?

When an individual self-injures this can have a major impact on their life and the lives of their family. It can cause a lot of distress and can be very difficult for professionals to treat effectively. If we can find out more about self-injury in 1p36 deletion syndrome, then we can provide parents with more information. This information will also help professionals to prevent and treat self-injury early. Self-injury has also been found to be high in people with several other rare syndromes and we wanted to see how 1p36 deletion syndrome compared with these. We also know from previous research that there are some behaviours that can predict self-injury, such as hyperactivity (always being 'on the go'), impulsivity (acting quickly without thinking) and repetitive behaviours (doing things over and over). We wanted to find out whether these behaviours are linked to self-injury in people with 1p36 deletion syndrome.

What did we do?

We sent out a questionnaire pack to the families of 54 individuals with 1p36 deletion syndrome. The questionnaires asked about several behaviours including self-injury, physical aggression, mood, hyperactivity and repetitive behaviour. They also asked about levels of ability. We also observed six children with 1p36 who were reported to show self-injury or aggression. This was to help us to understand what triggers certain problem behaviours. We were interested in two triggers in particular: the amount of attention being given to the child by an adult and whether they were being asked to do a task that they would find demanding or difficult. These things have been found to be linked to self-injury in people with other rare syndromes.

What did we find?

Twenty-three families sent the questionnaires back to us, the average age was seven and the majority (70%) were girls. The results from the questionnaires told us that ten (44%) children with 1p36 deletion syndrome were fully mobile, eight (35%) had normal vision, fourteen (61%) had normal hearing, four (17%) had some self-help skills and nine (39%) showed some speech. Fourteen (61%) children showed self injury and twelve (52%) showed physical aggression. Self-biting was found to be the most common form of self-injury. We compared these results from the same survey carried out with people from three other syndrome groups. We found that the number of children showing self-injury and aggression in the 1p36 deletion syndrome group did not differ from the number in the other syndrome groups. We also found that children with 1p36 deletion syndrome who self-injured were more likely to be very active and show stereotyped behaviour (such as repetitive movements). Children who showed aggression were more likely to be impulsive (act without thinking) and show repetitive behaviour (doing things over and over). Four of the six children we observed showed self injury in the form of biting or sucking their hands. In three of these children we found that their self-injury was related to the amount of attention they gained from an adult. We also found that some of the children showed high levels of smiling and laughing. Also, all of the children approached and moved away from us as different times.

What does all of this mean?

The findings tell us that although self-injury in children with 1p36 deletion syndrome is common, it is no more common than in a number of other rare syndromes. We also know that in children with 1p36 deletion syndrome, self-injury and aggression are linked to the same

sorts of behaviours as in other syndromes (being very active, impulsive and showing repetitive behaviour and movements).

We were interested to find that the children we observed were able to move towards and away from an adult. This indicated to us that they were showing signs of communication. This is very important for the treatment of self injury, as sometimes these signs of communication can later develop into self-injury. If we can work with children on these communication behaviours, then we may be able to prevent self-injury.

Appendix 1

Ethical Approval from the School of Psychology, University of Birmingham

UNIVERSITY OF BIRMINGHAM

Chair, School Ethics Committee

School of Psychology

August 26, 2008

App 06/08

Dear Abby:

Many thanks for your application to the School of Psychology ethics committee. Members of the committee have now commented on your application and have raised only one minor concern. The study proposes to assess excessive eating/appetite among other behaviours, however, we could not find this referred to in the information sheet

This item is minor and I am happy to provide Chairman's action for approval. Please do bear in mind the following points as you conduct your study:

1. Ethical conduct of the study remains your responsibility. Once investigations begin unexpected issues can arise and you are encouraged to think again about the situation of your participants.

2. You are free to make modifications to your procedures, without further ethical review, so long as you remain confident that your new procedures do not raise any general ethical problems or particular violations of BPS guidelines.

3. If funding is provided by the ESRC then outside review is mandatory from January 1, 2006. A Birmingham wide REC has been created for this purpose and applications can now be accepted by the Chairperson. There is a dedicated website for submissions.

If you have any questions or further issues arising then do not hesitate to contact me. I wish you the best of luck with your research.

Sincerely yours,

Appendix 2

Covering letter for carers

RARE CHROMOSOME DISORDER SUPPORT GROUP

Unique ...but not alone

UNIVERSITY OF BIRMINGHAM

School of Psychology

Edgbaston

Birmingham

B15 2TT

Project Director: Professor

Tel:

Date

Dear……

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 and for affected families. Briefly, the research is a study looking at different behaviours and cognitive impairments in children and adults with Mononsomy 1p36 Deletion Syndrome, for example memory, attention and concentration problems; and also exploring parents' experience of receiving a diagnosis of 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 at the above address or on.

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 Unique newsletter. In addition, the results of this research project may be published in a scientific journal.

If you wish to take part please complete a consent form and return this 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

Information Officer

Professor of Clinical Psychology

Unique

Appendix 3

Participant Information Sheet

Behavioural and Developmental Characteristics of Individuals with Monosomy 1p36 Deletion Syndrome.

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 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/guardians 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.

In addition, we will be seeking parents'/guardians' experiences of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome for their child. There is currently very little information available to parents when they receive this diagnosis. We would therefore like to conduct interviews with the principal caregiver.

What does the study involve?
You will be invited along to Birmingham University for the day to meet other families with individuals with Monosomy 1p36 Deletion Syndrome. This day will provide an information session for participants, families and Unique about behavioural phenotypes and the research conducted by the University, and a 'get together' for the members of the support group.

Following this day, we would like to visit individuals at their home/day placement/school etc in order to carry out some observations of feeding/eating difficulties and self-injury We will

visit your child/person you care for at their day placement for the day. During this time, we will carry out short observations of your child/person you care for in different social situations and during a series of games and activities. Video recordings of the observation sessions will be made, as it is necessary for another psychologist at the University of Birmingham to check the accuracy of the observations (additional information on videoing is provided further on in this information sheet). The different social situations and activities will be presented to your child/person you care for by two members of the research team. We will use three different social situations which will last 10 minutes each. The first situation will provide your child/ person you care for with lots of attention and we will play games with them. In the second situation we will not initiate any interaction with your child but we will interact if the child attempts to initiate interaction. In the third condition we aim to see how your child responds if we do not interact with them socially.

During our time at the day placement of your child/person you care for we will also carry out some observations to help understand what triggers certain problem behaviours. We will carry out observations as your child takes part in situations where levels are adult attention and demands are varied. The situations include three different conditions (10 minutes each), which your child will experience regularly in their normal day environment. The first situation is a "high attention" in which the researcher will interact with your child while they play with a preferred toy or game. The second condition is a "high demand" condition in which the researcher will ask your child to take part in a less preferred task and will continue to prompt and guide your child throughout the task. The final condition is a "low attention" condition in which your child will again have access to a preferred game or toy but this time the researcher will move their attention away from your child and will talk to the researcher. It is possible that these situations will cause an increase or decrease in particular behaviours. If your child becomes extremely distressed or is at excessive risk of injuring themselves we will immediately stop the session.

During the home visit, we may also conduct some short observations of your child/ person you care for within the home. This will be very similar to the method described above, only this time we will ask *you* to interact with your child/person you care for. The situations will be very natural and will be situations that are part of the regular routine of your child/person you care for. The situations will be similar to the high attention, low attention, and demand conditions conducted during the school assessments (see above) and we will ask you to think of a time in your daily routine where this situation may occur (e.g. high attention may occur when you play a game at a certain point during the day). We will then ask you to run through this situation so that we can observe any changes in behaviour that might occur during that time. In some cases it may be necessary to ask you to think of more specific situations in which self-injurious or aggressive behaviour usually occur and include this situation in the observations (for example it may be the case that you observe higher rates of these behaviours when a preferred activity is terminated. Therefore, we would include this situation in the observations). There may also be times when we ask you respond to your child/person you care for in a different way to that which you would normally do (e.g. during a low attention condition, we may ask you to turn around and talk to your child/person you care for briefly when behaviour occurs, rather than not paying any attention to the behaviour). Again, this is to observe any changes in behaviour that occur as a result of these situations and responses. It is possible that these situations will cause an increase or decrease in particular behaviours. If your child becomes extremely distressed or is at excessive risk of injuring themselves we will immediately stop the session.

Whilst at the day placement of your child/person you care for, we will also carry out some simple cognitive assessments to give us information about specific problems which may be faced by individuals with Monosomy 1p36 Deletion Syndrome, for example memory, attention and concentration problems.

In order to investigate parents'/guardians' experiences of receiving a diagnosis for their child you will be invited to attend an interview which will last approximately one and a half hours. This interview will be audio taped, and then transcribed and analysed by one of the researchers. Any quotes that are pertinent to the study will be included within the final publication, however all attempts to make such quotes anonymous will be made, and individuals will not be made identifiable. Where possible, these interviews will take place, whilst other members of the research team are conducting cognitive assessments or observations.

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 possible that parents/guardians may become distressed due to the nature of the questions asked during the interview. Every step will be taken to make sure that any distress is kept to a minimum. It is anticipated that parents/guardians will find the interview to be a positive experience, and will benefit from sharing their experiences, with the knowledge that this research could shape further research relating to their child's condition.

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 Unique, 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 Unique newsletter. Any requests for advice concerning your child/ the person you care for will be referred to Professor, Clinical Psychologist. It is possible that you may be invited to participate in further research after the study but consenting to participate in this study does not mean that you are obliged to do so.

Review:
If you have any concerns about the conduct of this study please contact Dr 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.

Appendix 4

Consent Form

Behavioural and Developmental Characteristics of Individuals with 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 the involvement of my child/the person I care for 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……...

Appendix 5

Questionnaire pack:

Background Information

Wessex Scale

Challenging Behaviour Questionnaire (CBQ)

Mood, Interest and Pleasure Questionnaire- Short-form (MIPQ-S)

The Social Communication Questionnaire: Lifetime (SCQ-L)

The Activity Questionnaire (TAQ)

Repetitive Behaviour Questionnaire (RBQ)

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?

_____

_____

_____

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 ☐

Please check your answers and go on to the next questionnaire.

THE 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 ☐

Please check your answers and go on to the next questionnaire.

THE MOOD, INTEREST AND PLEASURE QUESTIONNAIRE –

SHORT FORM (MIPQ-S)

Instructions:

- 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

*<u>engaged in activities</u>: i.e. when someone is actively involved in any activity such as a mealtime, social interaction, self-care task or social outing etc.

*<u>facial expressions</u>: 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):

Please check your answers and go on to the next 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

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 frustrated if left in one position for too long?**	**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**

Please check your answers and go on to the next questionnaire.

THE 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**

	Never	Once a month	Once a week	Once a day	More than once a day
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**

	Never	Once a month	Once a week	Once a day	More than once a day
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**
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**

	Never	Once a month	Once a week	Once a day	More than once a day
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**

Please check your answers and go on to the next questionnaire