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Adjustment and well-being among parents of children and adults with intellectual disabilities : Aetiology and behavioural phenotypes

Griffith, Gemma

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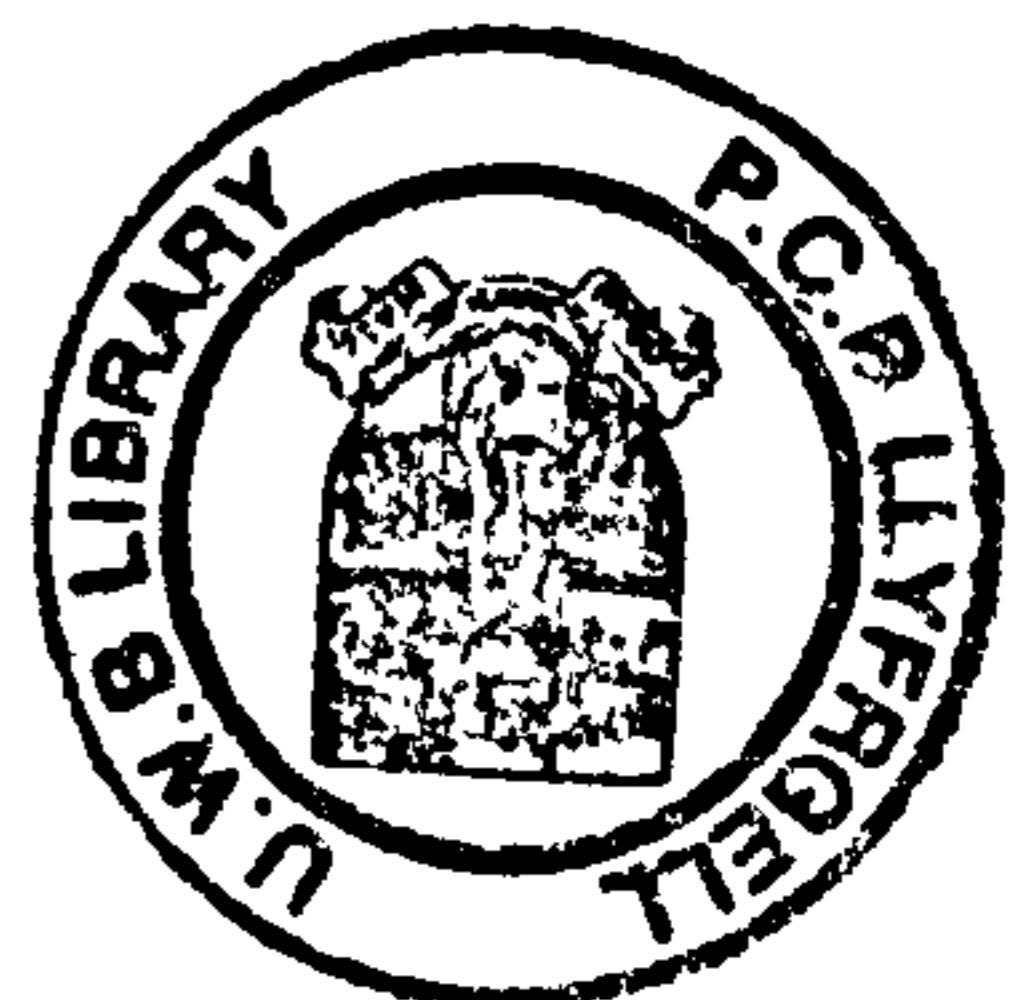
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Bangor University

**Adjustment and well-being among parents of children and adults with
intellectual disabilities: Aetiology and behavioural phenotypes.**

Gemma Maria Griffith

Thesis submitted to the School of Psychology, Bangor University, in partial
fulfilment for the degree of Doctor of Philosophy.

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Summary

Parenting a child with intellectual disabilities is a complicated experience, and parents of children with an intellectual disability often report more stress than parents of typically developing children. This thesis attempts to expand the existing knowledge base on parental adjustment in several ways; by using both quantitative and qualitative research approaches, examining issues surrounding the rareness of a syndrome, and by exploring a wide range of parent and child variables. This thesis is primarily about families of children with rare genetic syndromes, with the exception of Chapter 2, which examines parents of children with autism, Down syndrome, and mixed aetiology intellectual disabilities.

In Chapter 1, the existing literature on adjustment in families with a child with a rare genetic syndrome was critically discussed and recommendations for future research were made. In Study 1 (Chapter 2) the use of closely matched groups resulted in few differences found between aetiology groups on both child and maternal outcomes. In study 2 (Chapter 3) parents of children with rare syndromes who displayed challenging behaviour at least once a day were found to report high levels of stress, comparable to parents of children with autism. Mothers of adults with rare syndromes were the focus of a qualitative study (Chapter 4) in which it was found that mothers were heavily involved in maintaining adequate social and medical services for their offspring, and the strain this placed on them. In the final empirical study (Chapter 5) a multiple regression analysis on a large group of mothers of children with rare syndromes revealed child behaviour was not often predictive of negative or positive maternal measures, but child positive mood was.

Finally, in Chapter 6, findings from the four empirical studies were discussed in relation to their theoretical and methodological value, specifically with recommendations to include a wider range of independent and dependent variables within this area of research so as to better anticipate parental adjustment. Implications for future research and interventions are also discussed.

Chapter 1. Introduction: A review of the literature on adjustment in families of children with rare syndromes.

Chapter 1

When a child with an intellectual disability is born into a family, parents face a period of adjustment and adaptation to their child which continues throughout their lives. Some families adapt well to a child with an intellectual disability, creating meaning from their experiences and taking great pleasure in their child's development and achievements (Kearney & Griffin, 2001). Other families find the challenge of raising a child with intellectual disabilities outweighs their ability to cope, and show clinical levels of psychopathology (e.g., Olsson & Hwang, 2001; Richman, Belmont, Kim, Slavin, & Hayner, in press). This life-long process is a complex phenomenon, and much research has been devoted to the matter of why families vary so widely in their adjustment to having a child with an intellectual disability.

There is substantial evidence that the aetiology of a child's disability is an important variable in parental stress (e.g., Hodapp & Dykens, 2001; Hodapp, Wijma, & Masino, 1997). Although there has been much research on family adaptation to the more common conditions associated with intellectual disability, such as Down syndrome and autism, (Abbeduto et al., 2004; Holroyd & McArthur, 1976; Kasari and Sigman, 1997; Olsson & Hwang, 2001; Pisula, 2007; Ricci & Hodapp, 2003; Sanders & Morgan, 1997), little focus has been given to rarer conditions, such as children with rare genetic syndromes.

Behavioural phenotypes

Children with rare genetic syndromes are of interest to researchers because genetic syndromes are often associated with distinct behavioural patterns. These are known as 'behavioural phenotypes.' The phenotype of a syndrome reflects the increased likelihood of finding particular behavioural characteristics within a syndrome, although not every child with a given syndrome will show all

characteristics of the behavioural phenotype (Dykens, 1995). The behavioural phenotype may have rather specific aspects to it, for example, many individuals with Williams syndrome are prone to having many fears (Dykens, 2003).

Behavioural phenotypes have also been described as ‘direct effects’ (Hodapp, 1997), because the genetic make-up of the individual directly affects their behaviour.

A particular behavioural phenotype may also lead to ‘indirect effects,’ for example, if a child with a rare genetic syndrome is pre-disposed to exhibiting high (or low) frequencies of maladaptive behaviour and high (or low) levels of sociability, then those around them may feel and behave differently towards the child. Thus, indirect effects refer to the way a child’s behavioural phenotype influences their environment via their interactions with people around them (Hodapp, 1997).

The purpose of this Chapter is to draw together the available published research exploring adjustment in families who have a child with a rare genetic syndrome. This is in order to help highlight the methodological challenges faced by researchers in this area, and to use this previous research to help highlight any strengths and/or weaknesses in the research area, and to help inform the investigations conducted in this thesis. The methodological and conceptual challenges facing researchers studying adjustment among families with a child with a rare genetic syndrome will also be discussed. The thesis structure, and background to the empirical studies presented will be described towards the end of the Chapter.

Methodology

An extensive search was conducted electronically using the databases PsychINFO, Medline, and the Web of Science. The terms used for searches included the names of 80 rare genetic syndromes, drawn from the websites of Society for the Study of Behavioural Phenotypes and UNIQUE (the rare chromosome disorder

support group), and the terms famil*, maternal, paternal, sibling*, mother*, father*, relatives, stress, and adjustment. Data bases were searched for all years available. The inclusion criteria were that the syndrome had to be: (1) caused by a genetic deletion or abnormality, (2) focused on children or adolescents, (3) associated with intellectual disability, and (4) classified as rare (i.e. a condition which affects five or fewer individuals in every 10,000: Chatzimarkakis, 2009). Down syndrome was not included in the search criteria as it is the most common genetic syndrome, and there is already substantial research on families of individuals with Down syndrome (e.g., Hodapp, Ly, Fidler, & Ricci, 2003; Ricci & Hodapp, 2003). Research papers had to include data about current family functioning with either parents or siblings of a child with a rare genetic syndrome. The abstracts from all promising articles were reviewed, and the reference sections from articles obtained were searched to find any other relevant studies. Twenty-eight studies were found which met the criteria, twenty-six quantitative studies which were based on parental responses to questionnaires, and two qualitative studies.

Family adjustment in rare syndromes

As the research on parents of children with rare genetic syndromes is diverse with no underlying methodologies or concepts drawing the research together, the studies are presented and discussed according to child aetiological group.

We could find just 13 rare syndromes in which family functioning has been explored: 22q11.2 deletion, 4q, Alpert, Angelman, Cornelia de Lange, Cri du Chat, fragile X, Joubert, Prader-Willi, Rett, Smith-Magenis syndromes, Tuberous Sclerosis Complex, and Williams syndrome. All available studies on these syndromes will be described and discussed in turn. The gender of the caregivers was not reported in a number of these studies (although the vast majority of participants were described as

mothers). Therefore, the term 'parents' will be used when the exact gender distribution among primary caregivers is unknown.

22q11.2 deletion syndrome

22q11.2 deletion syndrome is the most frequent microdeletion syndrome, and it is estimated that around 40-50% of individuals with the syndrome also have an intellectual disability (Swillen et al., 1997). Just two studies from Germany were found, the first examined child behaviour and parental well being in infants (Briegel, Schneider, & Schwab, 2006), and the second focused on older children and adolescents (Briegel, Schneider, & Schwab, 2008).

In the earliest study, 22 parents (21 mothers, 1 father) of young children (aged 1 year 6 months to 3 years 11 months) participated. It was found that the behaviour problems of infants with 22q11.2 deletion syndrome were just slightly elevated when compared to the normative population, and that parents reported similar stress levels to the normative population. Although an association was found between child behaviour problems and maternal outcomes, both the children and parents in this study were very well adjusted (Briegel et al., 2006).

When examining behaviour in older children (aged 4 years to 16 years 11 months) with 22q11.2 deletion syndrome ($n = 77$), the picture changes somewhat. Briegel et al., (2008) found that 45% of children had a behaviour problem within the clinical range, which was associated with the severity of the child's intellectual disability. Maternal stress was higher compared to the normative population, and was correlated with higher child problem behaviours. Although these studies are cross-sectional, it appears that mothers of children with 22q11.2 deletion syndrome

are affected by the behaviour problems of their child, and feel more stress as their child gets older.

When interpreting these results, some methodological limitations should be borne in mind. Firstly, the intelligence levels of the children were estimated by the parents, and were not subject to a standardized test, Secondly, 38% of children in the first study and 29% in the second were reported as not having an intellectual disability. It would be interesting to see what the results would have been if the children were split into intellectual disability and non- intellectual disability groups. Finally, longitudinal studies are needed to further investigate the current finding as to whether parents report more stress and their child ages.

4q syndrome

4q syndrome is associated with mild to severe intellectual disabilities, and common behavioural issues include aggressive behaviour and hyperactivity (Strehle & Middlemiss, 2007). In the only study on parents of children with 4q syndrome, there was a single, open-ended question on family functioning; “How has your child contributed most to your lives?” The vast majority of parents (86%) described the positive contributions the child with 4q-syndrome had made, such as teaching patience, and a reminder of what is important in life. However, Strehle and Middlemiss (2007) do not explain how the remaining 14% of parents responded to the question. The overall positive response to the question highlights that positive aspects of having a child with a rare syndrome are often ignored in research, and yet are very salient to parents. This study did not explore any other aspects of family functioning, and thus the research on parents of children with 4q syndrome is very much in its infancy.

Angelman syndrome

The severity of intellectual disability in Angelman syndrome ranges from moderate to profound (Clarke & Marston, 2000), and frequent smiling and laughing, hyperactivity, and sleep disorder are common behavioural features of the syndrome (Clayton-Smith & Laan, 2003; Horsler & Oliver, 2006).

In the only study on parents of children with Angelman syndrome, van den Borne et al., (1999) compared mothers and fathers of children with Prader-Willi syndrome (n = 34) and Angelman syndrome together (n = 22: Mean age 7.25 years). Parental depression, self-esteem, and coping strategies were examined. Parents of children with Angelman syndrome reported higher self-esteem but also a higher sense of loss of control (e.g., feeling as though their hands are tied). Coping strategies and depression levels were similar to parents of children with Prader-Willi syndrome, although mothers reported slightly higher levels of depression than fathers. Unusually for family adjustment research, no child behavioural measures were included, and therefore we do not know whether child characteristics have any influence on parental outcomes. Furthermore, the results were not compared to other well researched aetiologies, thus it is difficult to get a sense of perspective on how parents of children with Angelman syndrome are coping from this single study

Apert syndrome

Apert syndrome is a rare genetic disorder that is characterized by severe craniosynostosis, caused by the premature fusion of skull bones. IQ levels range from normal to severe intellectual disability. In the only study on parental stress in Apert syndrome (Sarimski, 1998), 88% of children had an intellectual disability. The study used data from mothers and fathers from 41 families (41 mothers, 32 fathers). It was

found that the majority of children were psychosocially well adjusted and displayed low levels of aggressive and destructive behaviours, although older children were rated as being more demanding.

Around 25% of mothers reported stress levels in the clinical domain, and high levels of stress were related to child demandingness and acceptance problems (the author posited the latter was due to issues surrounding acceptance of their child's facial disfigurement). Mothers and fathers were more exhausted and socially isolated when compared to the normative population, although depression levels were similar to the normative population. There were no significant differences on ratings of well-being between mothers and fathers. In the second part of the study, Sarimski (1998) separated the children into two categories according to whether or not they had an intellectual disability. Mothers of children in the intellectual disability subgroup had more acceptance problems, but no other differences were found between the two groups, suggesting that whether a child had an intellectual disability or not was not a major contributing variable to parental stress.

From this single study it appears that parents of children with Apert syndrome are well-adjusted, as 75% did not report clinical levels of stress, and were reporting similar levels of depression to the normative population. Further research is warranted to validate this data. In particular, an interesting area of future research would be the association between parental stress and child facial appearance in Apert and other rare syndromes, particularly as some syndromes are associated with striking facial characteristics.

Cornelia de Lange syndrome

To date, just three studies on families of children with Cornelia de Lange syndrome have been conducted. The majority of individuals with Cornelia de Lange syndrome have profound or severe intellectual disabilities (Berney, Ireland & Burn, 1999). Common behavioural features include: anxiety, oversensitivity, sensory self-stimulation, self-injurious behaviour, and compulsivity (Basile, Villa, Selicorni, & Moltini, 2007).

In the earliest study Sarimski (1997) looked at 27 parents of children with Cornelia de Lange syndrome (mean age = 7.1 years). The primary focus of this study was on child communication and socio-emotional behaviours rather than problem behaviours. They found that child related parenting stress was higher than parents of typically developing children, and that lower child functioning and increased child age was related to higher parental stress. Little else was reported in this study, and although high frequencies of problem behaviours were reported (with around 40% of children showing self-injurious behaviours), the relationship between problem behaviours and parental outcomes was not thoroughly explored.

In a recent study in the Netherlands, Wulffaert et al., (2009) examined 37 parents of children and adults with Cornelia de Lange syndrome (range 1.4 years to 46.2 years: mean age 18 years). Parental stress was the only parental outcome measured and over a third of parents' stress levels reached the Parenting Stress Index's (PSI) cut-off point for 'very high'. Parenting stress was higher if their child had autism or more behaviour problems. However, with such a wide age range between individuals in a small sample, results should be interpreted with caution as there is likely to be much variability in life circumstances between these families due to the age differences in their offspring.

Richman et al., (in press) focused on child challenging behaviour and parental stress in children and young adults with Cornelia de Lange syndrome (n=25, age range 5.1-20 years) and Down syndrome (n=23, age range 5.1-24 years).

Parental stress was significantly higher in parents of children and young adults with Cornelia de Lange syndrome, with 40% of parents being above the 95th percentile for total stress on the PSI. Parental stress was associated with high levels of child self-injury, stereotypy, and lower levels of child pro-social and adaptive behaviour.

Although the research is limited and is based on small samples of parents, thus far it consistently shows that parents of children with Cornelia de Lange syndrome are at high risk for experiencing elevated stress, and this may be associated with the challenging behaviour displayed by the child. Given the high rate of self-injurious behaviours in this population (Basile et al., 2007), it may be useful in future research to determine precisely what types of challenging behaviour affect parental well-being.

Cri du chat syndrome

Cri du Chat syndrome is so named because of a characteristic 'cat-like' cry, apparent immediately after birth. The degree of intellectual disability ranges from profound to moderate (Cornish & Bramble, 2002; Sarimski, 2003) and self injurious behaviour and hyperactivity are associated with the syndrome (Cornish, Bramble, & Munir, 1998; Cornish & Bramble, 2002).

In the only study to date on families of children with Cri du Chat syndrome Hodapp et al., (1997) examined adjustment in both parents and siblings. They recruited 99 parents of children with Cri du Chat syndrome (91 mothers, 7 fathers, and 1 grandparent). The individuals with Cri du Chat syndrome were from 1 to 18

years old, with a mean age of 8.08 years. Parental stress was higher than reported by parents of children with mixed intellectual disability, and child problem behaviour was the strongest predictor of stress. Level of child adaptive behaviour was also a predictor of parental stress, but was not as strongly associated. Families reported receiving high levels of support, and total number of supporters was related to lower levels of parental stress.

The second part of the study was concerned with typically developing siblings, Hodapp (1997) also recruited 44 siblings (24 males, 20 females; mean age =11.2years) of children with Cri du Chat syndrome. It is the only study in this review to ask siblings directly about their perceptions. Parents and siblings were given slightly different versions of the Sibling Perception Questionnaire, one version measures sibling's feelings about the affected child's disability, and the other measures parent's perceptions of sibling's feelings. Parents felt that siblings communicated less when the child affected by Cri du Chat syndrome displayed more problem behaviours, and more overall stress within the family. Interestingly, parents perceived siblings being more affected by interpersonal concerns (such as parents not spending enough with the siblings) than reported by the siblings themselves. This study uses a comparatively large sample for rare syndrome family research, and finds that parents report more stress than parents of children of mixed aetiology intellectual disability.

Fragile X syndrome

Fragile X syndrome is the most common form of inherited intellectual disability, with an estimated occurrence of 1: 2000 - 4000 live births. Individuals may have mild to severe intellectual disability with associations with stereotypic

behaviours and social avoidance (Hall, Bernadis, & Ross, 2006). We found nine studies on families with children with fragile X syndrome. These will be discussed in turn, according to the methodology used: (1) Studies which used children with autism and Down syndrome as comparison groups, (2) Those studies which used other rare genetic syndromes as comparison groups, and (3) Those studies that examined parents of children with fragile X syndrome without the use of a comparison group will be explored.

Poehlmann, Clements, Abbeduto, and Farsad (2005) in the only qualitative study on this syndrome group interviewed mothers of children with fragile X syndrome (n=11) and Down syndrome (n=10). During the interviews, they explored reactions to the child's diagnosis, coping strategies, and mothers were also asked to describe their children. In terms of coping strategies, mothers of children with fragile X syndrome were more likely to engage in emotion-focused coping (e.g., wishful thinking). Both groups of mothers, however, were similarly positive in their descriptions of their children.

Two studies contrasted parents of adolescents with fragile X syndrome with adolescents with Down syndrome and autism (Abbeduto et al., 2004; Lewis et al., 2006), and are among the few studies to control for child characteristics or to match groups. Abbeduto et al., (2004) examined mothers of adolescents with a child with either fragile X syndrome (n = 22), Down syndrome (n = 39) or autism (n = 174). The three groups were similar in child age and socio-economic status, and differences in child gender and family size were statistically controlled for.

Mothers of adolescents with Down syndrome reported the highest levels of maternal well-being, mothers of adolescents with autism the lowest, with mothers of adolescents with fragile X in between. The strongest predictor of maternal well-

being was the severity of adolescent behavioural symptoms (as measured by the Autism Behaviour Checklist: ABC), the extent of which predicted maternal pessimism and fewer reports of reciprocated closeness.

In a smaller scale study, but with similar questionnaire measures to Abbeduto et al., 2004, Lewis et al., (2006) looked at mothers of adolescents with either fragile X syndrome (n=19) fragile X syndrome with a co-diagnosis of autism (n = 9), or Down syndrome (n=19). The participants were matched on IQ level. A co-diagnosis of autism was predictive of family conflict and less family closeness. In direct contrast to the findings of Abbeduto et al., (2004), adolescent challenging behaviour was not predictive of maternal well-being.

Three main factors must be considered when interpreting the data from the two latter studies. Firstly, the individuals with fragile X syndrome are older than in any other study in this review. Secondly, the ABC is designed to measure autistic-like behaviour and therefore it's validity for use with individuals with Down syndrome is questionable. Thirdly, the sample sizes are either very uneven between groups (Abbeduto et al., 2004), or were very small (Lewis et al., 2006).

Children with rare conditions were used as comparison groups in three studies on families with children with fragile X syndrome. Von Gontard et al., (2002) compared parents of boys with fragile X (n=49: mean age 8.2 years) to parents of boys with Spinal Muscular Atrophy (n=46: mean age 12.7 years) and a control sample of typically developing children. Parents of boys with fragile X syndrome reported higher stress levels than the other two groups, and the behaviour problems of the child were associated with higher parental stress.

Two studies published around the same time (although conducted by research groups in different countries) compared parents of children with fragile X syndrome

to both Prader-Willi and Williams syndrome (Sarimski, 1997; Van Lieshout et al., 1998). Sarimski (1997) compared child behaviour and parental outcomes in three rare genetic syndromes (age range 1-12 years old), fragile X (n=30), Prader-Willi syndrome (n=35) and Williams syndrome (n=35). They found that the average score for child behaviour problems was within the clinical range, and were at similarly high levels across all three syndrome groups. Child and parent-related stress were also similarly high across the syndromes. Sarimski (1997) concluded that type of syndrome had less of an impact on maternal outcomes than factors such as maternal coping resources and social support. However, this may be because the impact of child behaviour on parents was not examined, which is often the strongest predictor of maternal outcomes.

Van Lieshout et al., (1998) looked at the same rare syndromes as Sarimski (1997) although used a very different approach which focused on the central orientation of child personality rather than child challenging behaviour. Groups of parents of children with fragile X (n=32: mean age 10.5 years) Prader-Willi syndrome (n=39: mean age 10 years) and Williams syndrome (n=28: mean age 9.4 years) were used in this study. Van Lieshout et al., (1998) examined the wider family context and the interrelations between variables, such as parental behaviour (such as warmth, anger, and limit-setting), and the wider family context (such as parental consistency, family stress and marital relationships).

Significant differences in personality were found between the syndrome groups, children with fragile X syndrome were rated as being lower in agreeableness than children with Williams syndrome, and lower in conscientiousness than children with Prader-Willi syndrome. Although this study highlights the importance of taking wider family dynamics into account, it failed to examine the child characteristic

strongly associated with parental stress; child problem behaviour, and thus could be missing the strongest associations important to family adjustment.

Lastly, we found three papers which examined within-group characteristics of fragile X syndrome (Johnston et al., 2003; McCarthy et al., 2005; Hall, Burns, & Reiss 2007). Johnston et al., (2003) examined 75 mothers with a child with fragile X syndrome (mean age = 10.9 years). Maternal stress levels were significantly higher in the child related stress domain (which looks at how the child's behaviour impacts on the parent) than in the normative population, but were within the normative range in the parent domain (to what degree are parents able to cope with parenting). Additionally, child behaviour problems were significantly associated with higher overall maternal stress and lower feelings of competence. Neither the child's age nor intelligence level was associated with maternal stress.

McCarthy et al., (2005) analyzed Australian mother and father outcomes separately in 40 families who had a child with fragile X syndrome (mean age = 10.4 years). Both mother and father ratings of their child's problem behaviour were high, and the average score was within the 'at risk' category for problem behaviours. Although child behaviour was not predictive of maternal stress, it did predict paternal well-being, suggesting that mothers and fathers may be affected differently by their child's problem behaviour.

Hall et al., (2007), in the largest study on families with a child with fragile X syndrome to date (n =150 family quartets, including both biological parents, child with fragile X syndrome: mean age=10.9 years, and an unaffected sibling: mean age=11.4 years), examined wider family dynamics. Three main findings were reported. Firstly, child IQ did not have an effect on maternal distress. Secondly, both the child with fragile X syndrome and their unaffected siblings had equal effect on

maternal distress. Finally, maternal distress did not appear to influence child behaviour problems of either child.

The two studies that examined the wider family context among families of children with fragile X syndrome (Hall et al., 2007; Van Lieshout et al., 1998), highlighted the importance of doing so in order to avoid making false relations between two variables, e.g., that the child with fragile X is the main cause of parental stress when typically developing siblings have an equal influence (Hall et al., 2007). Although the studies discussed are all on families of children with fragile X syndrome, the results are not consistent. Of the six studies that examined the relationship between child behaviour problems and parental stress, four found that child behaviour problems were significantly associated with higher overall parental stress (Abbeduto et al., 2004; Johnston et al., 2003; Hall et al., 2007; Von Gontard et al., 2002), whereas two found no such relationship (Lewis et al., 2006; McCarthy et al., 2005). Perhaps the numerous approaches to this subject may underlie the variability in results.

Joubert syndrome

Joubert syndrome is a rare neurogenetic disorder, and the severity of intellectual impairment varies between individuals; some children are only mildly cognitively affected while others have severe intellectual disability. Luescher, Dede, Gitten, Fennell, and Bernard (1999) examined 49 primary caregivers of children with Joubert syndrome and measured child developmental problems along with parental outcomes such as depression, parental coping style, strain, and family function. They found that child impairment level was not related to parental burden, but was more dependent on parental coping strategies and level of family

functioning. Luescher et al, (1999) asserted that this showed that parental functioning was determined by the parent's ability to adjust and cope with Joubert syndrome, rather than the difficulties associated with the syndrome itself (similar to Sarimski, 1997). Given the lack of any other studies on Joubert syndrome, and no comparison groups used, it is difficult to draw any definite conclusions on family functioning in this population

Prader-Willi syndrome

Typically, individuals with Prader-Willi syndrome are classed as having a mild intellectual disability; and common behavioural problems include obsessive compulsive behaviours and temper tantrums. (Dykens, Hodapp, Walsh & Nash, 1992). Four studies about parents with children with Prader-Willi syndrome were found, three between-group studies and one focusing on Prader-Willi syndrome alone.

In a within-group study, Hodapp et al., (1997) recruited forty-two parents (39 mothers and 3 fathers) who had children between the ages of 3 and 16 years old (mean age = 10.3 years) with Prader-Willi syndrome. Parental stress, support, and child characteristics were explored. It was found that parental stress and pessimism were the prevalent negative outcomes, and child problem behaviour was predictive of parental stress. In particular, the strongest predictor of parental stress was behaviours characteristic of Prader-Willi syndrome; overeating and skin-picking. This finding demonstrates how important it is to include syndrome-specific behavioural measures that are salient to the syndrome being investigated, as it was not explored in any other studies on Prader-Willi syndrome.

All of the three studies using a between-group approach have been described earlier in this chapter, thus only the main findings as related to parents of children with Prader-Willi syndrome will be reported. All the studies found that stress levels of parents of children with Prader-Willi syndrome are similar to those experienced by other parents of children with rare genetic syndromes, including Cri du Chat, Smith-Magenis, Williams, and fragile X syndrome (Hodapp et al., 1998; Hodapp et al., 1997a; Sarimski, 1997).

Additionally, Sarimski (1997) found that the aetiology of the child was not predictive of parental outcomes (see page 14 for details). Van Lieshout et al., (1998: see page 15 for details) found that children with Prader-Willi syndrome were significantly more conscientious (possibly due to higher cognitive ability), and higher on irritability than children with Williams and fragile X syndrome. Parents of children with Prader-Willi syndrome showed significantly more parental anger than the other two groups of parents, which was related to lower conscientiousness and lower openness in their children. This is the only study on Prader-Willi syndrome to specifically look at parental anger, and is suggestive of complex family dynamics between child characteristics and parental outcomes.

In the most recent study on Prader-Willi syndrome, van den Borne et al., (1999) found that depression levels and coping strategies were similar to parents of children with Angelman and Prader-Willi syndrome (see page 5 for details on the study). There were some significant differences between the two groups; parents of children with Prader-Willi syndrome reported lower self-esteem but not as much loss control (i.e. not feeling as able to handle their affairs as well as before their child was born) then parents of children with Angelman syndrome.

All research on parents of children with Prader-Willi syndrome suggest that parents have similar stress levels to parents of children with other rare syndromes, and some further suggest that aspects of the behavioural phenotype of Prader-Willi syndrome is likely to contribute to this (Hodapp, 1997; van Lieshout, 1998).

Rett syndrome

Rett Syndrome is a severe neurodevelopmental disorder which mainly affects girls, and is associated with severe intellectual disability and physical disabilities (Laurvick et al., 2006). Three studies examining family functioning were found, two on parents and one on typically developing siblings (Laurvick et al., 2006; Mulroy, Robertson, Aiberti, Leonard, & Bower, 2008; Perry, Sarlo-McGarvey, & Factor, 1992).

Perry et al., (1992) examined both mothers and fathers of girls with Rett syndrome (n=29 families), the girl's ages ranged from 2 to 19 years (mean age=9 years 5 months). Both mothers and fathers reported higher parent-related stress than the normative population, with 23-31% being classed within the clinical range for stress. The adaptive functioning of the child was generally low, and was not related to parental outcomes.

In a much larger study (n=135) Laurvick et al., (2006) looked at the physical and mental health of mothers caring for a child or adult with Rett syndrome (age range 3 -27 years; mean 12.5 years). Both the physical and mental health of mothers of children with Rett syndrome was lower than the normative population. Like Perry et al., (1992) child adaptive behaviour was not related to maternal mental health. However, unusual facial movements (which is a behaviour associated with Rett syndrome), were strongly associated.

In a large qualitative study on siblings, Mulroy et al., (2008) asked parents of children with Rett syndrome (n=141) and Down syndrome (n=186) to state whether they felt any benefits or disadvantages of their child with disabilities on their typically developing siblings. Among parents of children with Rett syndrome, 82% of parents reported disadvantages, and 71% reported benefits, which was very similar to parents of children with Down syndrome (75% reporting a disadvantage and 80% reporting benefits). There were no substantial differences in parental response, with the exception of parents of children with Rett syndrome, who felt the family as a whole were affected by their child's preference for routine. Both parental groups reported very similar benefits to the sibling such as increased maturity, a more caring attitude, and increased tolerance toward other people.

The available literature on family functioning in Rett syndrome is limited to these three studies, we know that parents report more stress than the normative population, and that parental stress does not appear to be related to their child's level of adaptive function. This lack of association however, may be due to little variability in intellectual disability levels within the group, as many were classed as having severe intellectual disabilities. The studies also looked at a limited number of variables, and unlike most other research on rare genetic syndromes, did not examine the influence of child behaviour on parents.

Smith-Magenis syndrome

Smith-Magenis syndrome is associated with moderate intellectual disability, hyperactivity, aggressive outbursts, and a high degree of self-injurious behaviour (Greenberg et al., 1996). Two studies were found on Smith-Magenis syndrome; in a within-group study, Hodapp et al., (1998), examined child behaviour and parental

stress in 36 families (33 mothers and 3 fathers). The mean age of the children was 8.35 years, and a very high proportion of the children scored at or above clinical cut-off for problem behaviours (78%). The relationship of problem behaviours to parental outcomes in Smith-Magenis syndrome were not clear-cut, and child maladaptive behaviour was not associated with parental stress, although it was predictive of parental pessimism. The strongest predictor of parental stress was lower child socialisation scores.

In a between-group study, Fidler, Hodapp, and Dykens (2000) compared parents of children with Smith-Magenis syndrome to parents of children with Williams and Down syndrome. There were 20 children in each syndrome group, and all children were aged between 3 and 10 years. Statistical measures were taken to control for income, maternal age, and child gender, and these were not found to be significant co-variates. Children with Smith-Magenis syndrome displayed higher levels of maladaptive behaviour (80% reached clinical cut-off), than children with Down (40%) and Williams syndrome (75%). Parents reported significantly higher levels of pessimism and parent and family stress than parents of children with Down syndrome, and slightly higher than parents of children with Williams syndrome. Child problem behaviour was strongly correlated with parental stress, and the authors proposed that the behavioural phenotype of the child contributed to higher levels of family stress in Smith-Magenis and Williams syndrome.

In summary, Fidler et al., (2000) found that child problem behaviours were associated with maternal stress, while Hodapp et al., (1998), did not. Both studies used a similar methodology, and the same measure of child behaviour, and both reported that around 80% of children with Smith-Magenis syndrome reached clinical

cut off for problem behaviour. It is therefore unclear why this discrepancy occurred and thus more research is warranted to explore this.

Tuberous sclerosis complex

Kopp, Muzykewicz, Staley, Thiele, and Pulsifer (2008) conducted the only known study to date to examine stress among parents of children with Tuberous sclerosis complex (n=99). Tuberous sclerosis complex is a multisystemic genetic disorder which occurs in around 1:6000 live births (O'Callaghan & Osborne, 2000). Multiple organ systems such as the brain, lung, and heart are affected by hamartomatous growths, and 44-55% of individuals with Tuberous sclerosis complex have an intellectual disability. The children in the current study were aged 6 months to 17 years old, and 48.2% of the children had an intellectual disability. Forty percent of children were reported to have significantly elevated behaviour scores, and those with an intellectual disability were more likely to display problem behaviours. From these 99 families, 45 additionally completed questionnaires about their psychological well-being and parent-related stress, of these, around 50% reported clinically significant parenting stress, and this was associated with higher child problem behaviours and lower child IQ.

However, a major methodological issue is that no information is available on how the 45 parents were selected from the 99 families, therefore the demographic and child characteristics in that sample are unknown, for example we do not know how many of the children had an intellectual disability, therefore, results must be interpreted with caution.

Williams syndrome

Williams syndrome is associated with mild to severe levels of intellectual disability, and individuals with Williams syndrome tend to be highly sociable, but also display problem behaviours such as hyperactivity, anxiety, and fear (Dykens, 2003). There are three between-group studies which include a group of parents with children with Williams syndrome, all of which have been described previously in this review, therefore just the main findings relevant to Williams syndrome will be reported here.

Sarimski (1997, see page 14 for details) found that children with Williams syndrome had similarly high levels of behavioural problems, and their parents have similar levels of stress to parents of children with fragile X and Prader-Willi syndrome, and thus suggested that child aetiology did not predict parental outcomes. Van Lieshout et al., (1998, see page 15 for details), found that children with Williams syndrome were rated as higher in agreeableness than children with fragile X or Prader-Willi syndrome, and that fewer feelings of parental anger were reported by families with a child with Williams syndrome than in the other two syndromes. Overall though, there were no significant relations between family context variables, child personality characteristics and parental behaviour among families with children with Williams syndrome, although relations of this type were found among families with a child with fragile X and Prader-Willi syndrome.

This is in contrast to a later study which found that child characteristics *were* predictive of parental outcomes (Fidler et al., 2000, see page 22 for details). Parents of children with Williams syndrome reported similar levels of stress to parents of children with Smith-Magenis syndrome. Child problem behaviour and age (negative predictor) predicted 59% of parental stress.

Throughout the three studies, it is consistently reported that parental stress is similarly high in parents of children with Williams syndrome and parents of children with other rare syndromes. Unusually, only one out of the three found that stress was linked to child characteristics (Fidler et al., 2000). Further research is needed to determine why this is; is there something about the behavioural phenotype of Williams syndrome that has less of an impact on parental behaviour than in other rare genetic syndromes?

Discussion

The research presented in this review has been conducted by researchers in different countries, each using different approaches and instruments to measure child, demographic, and parental variables. Although it may be argued that a number of approaches to measurement are required in order to capture family functioning throughout diverse rare genetic syndrome groups, this approach has thus far not always resulted in consistent results, even between studies investigating the same rare genetic syndrome.

In the main, the only consistent findings concern parents of children with rare syndromes in relation to other well researched comparison groups. All studies (with the exception of Briegel et al., 2006) which compared parents of children with rare syndromes to normative data or parents of typically developing children, found that parents of children with rare syndromes reported higher levels of stress (Perry et al., 1992; Sarimski, 1997; 1998; Von Gontard et al., 2002). There is also consistent evidence to suggest that parents of children with rare syndromes report more stress than parents of children with mixed aetiology intellectual disability (Hodapp et al., 1997; Hodapp et al., 1998; Von Gontard et al., 2002) parents of children with Down

syndrome (Abbeduto et al., 2004; Fidler et al., 2000; Lewis et al., 2006; Richman et al., in press), but less stress than parents of children with autism (Abbeduto et al., 2004; Lewis et al., 2006).

Where the literature is less consistent is when examining child and demographic variables, and the influence of these on parental reports of stress and well-being. The majority of studies look at child problem or adaptive behaviour, and consistent with the wider family adjustment literature (e.g., Olsson & Hwang, 2001), finds a strong correlation between child problem behaviour and parental stress and depression (Briegel et al., 2008; Hodapp et al., 1997; Richman et al., in press; Wulffaert et al., 2009), however, some studies do not find this association (Lewis et al., 2006; McCarthy et al., 2006). The literature also shows that lower levels of child adaptive behaviour are associated with higher parental stress (Hodapp et al., 1997; Sarimski, 1997). No other child, demographic, or family level measures (such as marital satisfaction) have been consistently investigated as independent variables.

As the behavioural phenotype of each syndrome is unique, we can not easily compare results across syndromes. Additionally, due to the lack of methodological similarity of the approaches to assessing family adjustment in this population, it is equally problematic when attempting to bring together the literature about a single syndrome. Overall, the findings resulting from these varying methods of approach call for a rethink in how to address family adjustment in rare genetic syndromes.

One of the aims of this review was to examine the issues surrounding methodological approaches, and so the following issues will be discussed: (1) Issues surrounding the measurement of child characteristics, (2) Internal validity, (3) Use of family stress models in research with children with rare syndromes, and (4) Recruitment issues.

Measurement of child characteristics

The main difficulty in understanding the current literature is the lack of consistency in the type of child and demographic variables measured between studies, and also the instruments used to measure them vary widely between studies. An approach is needed in which the independent variables are measured using the same instruments - this would ensure a consistency currently lacking in the research literature.

There are additional difficulties with the validity of the measures; Instruments assessing adaptive or challenging behaviour are often not validated for children with rare genetic syndromes; for example, one measure which was used in seven of the reviewed studies is the Child Behaviour Checklist (CBCL) was not been designed for use with populations with severe/profound disabilities. This measure was designed to measure psychopathology, not behaviour problems, among typically developing children (Perrin, Stein, & Drotar, 1991). The CBCL contains items that are unlikely to be applicable to parents of children who have severe/profound intellectual disability, such as "Fears he/she might think or do something bad" (Achenbach & Edelbrock, 1983). In order to have an accurate assessment of child problem behaviours, measures need to be developed and used which are validated for use with children with rare syndromes.

Children with rare genetic syndromes are associated with unique behavioural phenotypes, yet only two of the studies reviewed in this chapter included syndrome-specific child behaviour measures when examining parental outcomes, both of which found that syndrome-specific behaviours were highly correlated with parental stress. Among parents of children with Rett syndrome, the extent of the child's unusual facial movements was strongly associated with parents' mental health (Laurvick et

al., 2006). In children with Prader-Willi syndrome, the behaviours of overeating and skin-picking were associated with parental stress (Hodapp et al., 1997). The presence of these unusual behavioural phenotypes may be particularly stressful for parents, and yet are seldom explored in family adjustment research. This is likely to be because there are no validated measures for the many types of unusual behaviours found in children with rare genetic syndromes (such as pulling out own toe and fingernails in Smith-Magenis syndrome: Greenberg et al., 1991). Although it is important to use a well validated measure, in doing so, unusual behaviour phenotypes may be missed, as well as the impact of these on parental well being.

Internal validity

The assumed hypothesis in the majority of the reviewed studies is that any differences among parents are the result of the behavioural phenotype of the child (Briegel et al., 2008; Hodapp et al., 1997). Yet there are numerous other factors that may account for such differences, including family socio-demographics, (e.g., income, marital status, socio-economic status) biological vulnerabilities, (mothers of children with fragile X syndrome are sometimes carriers of the permutation gene), and child characteristics (e.g., age, gender, severity of intellectual disability, behaviour problems). Some studies in this review do statistically control for some of these variables (Abbeduto et al., 2004; Lewis et al., 2006), but most do not.

Therefore, the internal validity of the research may be questionable. Replication of some of the existing research using matched samples and consistent measurements to evaluate both child characteristics and parental well-being may help provide the consistency which is currently lacking within the family literature surrounding rare genetic syndromes.

Recruitment issues

The vast majority of the reviewed studies (22/28) recruited participants exclusively from members of the relevant national or international syndrome parent support group. The remaining studies used samples recruited from a combination of parent support groups, national advertising, specialist hospitals, and by mailing recruitment leaflets to special education units or genetic clinics (Abbeduto et al., 2004; Johnston et al., 2003; Kopp et al., 2008; Lewis et al., 2006; Poehlmann et al., 2005), and one did not state how their sample was recruited (McCarthy et al., 2006). Some of these syndromes have only recently been identified, so parents who are in touch with the relevant national or international support group may represent a particularly pro-active, committed, and well informed sub-group of parents. Conversely, parents may approach support groups because their child may be more challenging and they may be seeking support (Finegan, 1998). Therefore, these studies may not reflect the larger populations of families of children with rare syndromes. There is no easy alternative to recruiting participants from parent support groups. Finding and contacting families via other means would be a considerable challenge for researchers, but efforts should be made to include parents who do not belong to a syndrome support group.

Use of family stress models in research with children with rare syndromes

It is important to bear in mind in which direction the field is developing as a whole. The use of between group studies are useful as a starting point in research about families of children with rare genetic syndromes, but finding differences between syndrome groups still does not provide an explanation for them, and specific questions still need to be addressed (e.g., to what extent does a behavioural phenotype of a syndrome contribute to parental psychosocial outcomes?).

In the wider family adjustment literature, attempts have been made to disentangle the complex interactions that operate within families. The more popular frameworks for analyzing parental stress include the Double ABCX model (McCubbin & Patterson, 1983), Lazarus and Folkman's (1984) Process model of Stress and Coping, and the Family Adjustment and Adaption Response model (FAAR: Patterson, 1983). A common element to these theories is the recognition that parental coping strategies play an important role in determining adjustment outcomes.

The Double ABCX model (McCubbin & Patterson, 1983) comprises the stressor element (A), such as demands of the child, and two mediating variables of resources for meeting the demands and needs (B), and the meaning the family assigns to the situation (C). Finally, the outcome variable is family crisis adaption (X). Family adaption is regarded as a continuum of outcomes, encompassing families who adjust well, and those that continuously struggle to cope. This model has not been explicitly used in research on parents of children with rare syndromes, although elements of it have been examined such as the influence of the behaviour of the child (demand) on maternal stress (Wulffaert et al., 2009).

The FAAR model (Patterson, 1998), consists of two elements: Family demands (stressors and strains) and family capabilities (practical and psychological resources and coping behaviours), when demands outweigh perceived capabilities, a crisis is reached and the family aims to restore balance by changing coping behaviours and/or gathering new resources to adapt to the situation. This model attempts to demonstrate how, when faced with a stressor, families attempt to balance problems and resources in order to preserve a typical level of family functioning.

None of the studies in this Chapter were solely based on family adaption models, or theoretical models of the variables used. For example, although three studies measured maternal coping (Abbeduto et al., 2004; Lewis et al., 2006; van den Bourne et al., 1999), just one explicitly related this to a theory of coping (van den Bourne et al., 1999). Lazarus and Folkman's (1984) theory of coping proposes that coping is a dynamic interaction between a person and their situation, in which a person tries to maintain a balance between their own resources and the demands of the situation. The appraisal of a situation, (e.g. the demands of a child with intellectual disabilities) depends on the characteristics of the child, their parents and family. Whether the situation is perceived as stressful depends on the parents' appraisal (primary appraisal) of their situation and anticipated consequences for family well-being. In doing so, parents will appraise the resources they have (secondary appraisal), and based on this appraisal process parents will usually apply a combination of problem and/or emotion-focused coping strategies. Although this is a model which has been widely used in family adjustment literature, the only reviewed study which referred to Lazarus and Folkman's (1984) theory of coping in their introduction (van den Bourne et al., 1999) did not relate their findings to the theory of coping in the discussion.

The majority of research on parents of children with rare genetic syndromes has focused on the relationship between child variables and maternal measures, and fails to consider the wider family context or existing models of family functioning. There is growing evidence that all members of the family are affected by a child with a rare syndrome, and thus fathers and siblings need to be taken into account as well as the mother (Hall et al., 2007; van Lieshout et al., 1998). Additionally, if the focus of research is solely on the mother/child relationship, researchers may be

missing some influential variables from the wider family context, such as the behaviour problems of typically developing siblings (Hall et al., 2007). Attempting to analyze family function models with reciprocal, bidirectional causal effects is a considerable challenge in the wider family literature (e.g., Saloviita, Italinna, & Leinonen, 2003), and even more so in families of children with rare syndromes because of the difficulties in recruiting large samples. Perhaps a more consistent focus on parental models of stress when designing studies on families with a child with a rare syndrome may result in the development of a more coherent area of research.

Summary

In conclusion, the literature area on families of children with rare genetic syndromes is still in its infancy, and much of the reviewed research is exploratory in nature. This has resulted in inconsistent findings, even on research on children with the same genetic syndrome. A clearer focus is needed on the methodological issues highlighted in order to make a stronger, more coherent literature base.

The majority of this thesis is based on data collected as part of a wider, collaborative project entitled the “Three Syndromes” project. This was a multi-site collaboration in conjunction with the University of Birmingham and the Institute of Psychiatry, London. The aims of the wider project were to:

1. Further describe the behavioural phenotypes of Cri du Chat, Cornelia de Lange, and Angelman syndromes.
2. Develop our understanding of the role of social/environmental variables in behaviour disorders associated with these three rare genetic syndromes.

3. Explore the factors that relate to both negative and positive adjustment in families (specifically, parents) of children and adults with rare genetic syndromes.

The researchers at the University of Birmingham and Institute of Psychiatry were concerned with aims no 1 and 2. The aim of the current thesis was to investigate the third aim and more specifically to:

1. Use a range of methodological approaches (including qualitative and quantitative research) to explore adjustment among families with children with rare syndromes and other aetiologies
2. Explore unique difficulties that parents of children with rare syndromes may experience, in particular, how the *rareness* of a syndrome may contribute to parental adjustment.
3. Be involved in the “Cross syndrome” study exploring negative and positive adjustment in parents of children across a variety of rare syndromes.
4. Examine a wide range of child characteristics which may contribute to parental stress, depression, and positive outcomes.
5. Use qualitative methods to investigate parents of adults with rare syndromes, with a specific focus on their experiences of support services for their offspring.

Chapters 3, 4, and 5 are a direct result of the Three Syndromes collaboration.

Chapter 2 is a secondary analysis of data held at Bangor University as a result of the ‘Special Needs and Families Research Project’ (SNFRP). The author was not involved in this SNFRP project, but used the resulting SPSS database to select the

matched groups and perform the subsequent statistical analyses. These data had not previously been analysed.

Chapter 3 uses the data collected during the wider Three Syndromes project. This involved research visits to 60 families throughout the UK who had a child with either Cri du Chat, Cornelia de Lange, or Angelman syndrome. Four researchers, including the author of this thesis, were involved in the data collection. Each research visit was conducted by two of the research team, and each visit lasted 1-2 days in order to conduct functional analysis assessments. The author was not involved with the data resulting from the functional analysis, which was used for a separate PhD project at the University of Birmingham. Only the data gathered from a family questionnaire pack given to parents was used in Chapter 3. The measures used in the family questionnaire pack were selected and formatted by the author of this thesis. These measures were then sent to parents before each research visit. Around two weeks prior to each visit, the author of this thesis also conducted Vineland Adaptive Behaviour Scales-Second edition interviews (VABS; Sparrow et al., 2005) via the telephone with all primary caregivers. The data from the family questionnaire pack was entered on to an SPSS database and analysed by the author. The results of the analysis of the family questionnaire and VABS-II data is presented in Chapter 3.

The qualitative study presented in Chapter 4 was not part of the initial Three Syndromes project but the idea arose thorough talking to families who participated in the Three Syndromes project about their experiences with social and medical services. It emerged that many had difficulties with statutory services and this caused some parents considerable strain. This study was primarily initiated, designed, conducted, and analysed by the author of this thesis.

The family data from the Cross syndrome project are presented in chapter 5. The Cross syndrome project is a longitudinal study (conducted by Chris Oliver at the University of Birmingham) of families of children with various rare syndromes, and is primarily concerned with the developmental trajectory of children with rare syndromes. There have been two previous 'waves' of questionnaires sent with at least a 3 year gap between each wave. For the third wave of the Cross syndrome project, the author of this thesis had the opportunity to add questionnaires examining family functioning to the existing Cross syndrome project. I selected and formatted the family and demographic measures in the questionnaire packs, and assisted in the data entry at the University of Birmingham. The presented data was analysed independently of the Cross syndrome study by the author of this thesis.

Additionally, as part of the dissemination of the findings from the Three Syndrome project, the Three Syndromes research team wrote and produced three separate informational DVD-ROMs for parents and professionals interested in Angelman, Cornelia de Lange, or Cri du Chat syndromes. The DVD-ROM had an explicit focus on challenging behaviour and information on strategies to help parents cope with these behaviours. It also included personal accounts from parents about their children and families. The author of this thesis conducted interviews with parents for this DVD-ROM and some quotes from these interviews have been included in the discussion (Chapter 6).

Structure of Thesis

Each of the Chapters takes a different methodological approach towards examining the question of whether the aetiology of a child or adult with an intellectual disability affects family functioning. The four empirical investigations reported in this thesis explore well-being among parents of children and adults with

rare syndromes, with the exception of Chapter 2, which uses groups of parents of children with Down syndrome, autism, and mixed aetiology intellectual disabilities. This second chapter is independent from the Three Syndromes project, and was a secondary analysis of a data set investigating parental adjustment to having a child with an intellectual disability. This Chapter intended to examine the methodological issue of whether differences found between various aetiology groups could be due to variables other than the aetiology of the child. From this large data set, we extracted three closely matched groups of children with Down syndrome, autism, and mixed aetiology intellectual disability in order to control for child variables. Statistical analysis revealed that despite matching the groups, children with autism were rated as having more problem behaviours and lower levels of social competence than children with Down syndrome and mixed aetiology intellectual disabilities. In terms of maternal outcomes, we found little evidence of group differences.

Chapter 3 was concerned with families of children with three rare syndromes that have seldom been examined, and was thus an exploratory study. Although the recommendations that emerged from the study in Chapter two was the use of matched groups, this was not feasible for parents with a child with a rare syndrome, due to the small sample sizes in this study. The data are taken from the “Three Syndromes” project and explore the well-being of parents of children with Cornelia de Lange (n=16), Cri du Chat (n=18), and Angelman syndrome (n=15). These data were compared to a matched group of parents of children with autism, extracted from a previous data set (n=20; Hastings, Beck, & Hill, 2005). Parents of children with Angelman syndrome consistently reported the highest levels of psychological distress, and parents of children with Cornelia de Lange the syndrome the lowest, with parents of children with Cri du Chat syndrome and autism scoring between

these two. Positive psychological functioning was independent of negative outcomes and was similar across the four aetiology groups.

The fourth chapter in this study examined parents of adults with either Cornelia de Lange, Cri du Chat or Angelman syndrome (n=8). The idea for this study arose from talking to parents of children and adults with these rare syndromes at family meetings and conferences, where the subject of difficulties with social services would often arise. As parenting an adult, or difficulties with social services were not examined in the data gathered by the Three Syndromes Project (presented in Chapter 3) or in previous research, we used qualitative methods to examine mothers' experiences of social and health services for their adult offspring. Four themes emerged from the thematic content analysis: (1) Uneven medical and social care service provision, (2) The inertia of social care services, (3) Mothers as advocates, and (4) The rarity of their offspring's syndrome. In particular, mothers reported undergoing substantial stress as a direct result of difficulties with accessing appropriate social or health services for their offspring. This study may help inform care service providers about how best to support young adults with rare genetic syndromes and their carers.

The final empirical study, using the data from the "Cross syndrome" project (Chapter 5), examined the question of to what extent child and demographic variables predict maternal positive and negative outcomes within a large mixed sample of children with rare genetic syndromes. This helps extend the current direction of methodological enquiry into examining a wider range of child variables, some of which may be particularly salient to research on children with rare genetic syndromes. The study looks at the well-being of large group of mothers of children with various rare syndromes. Regression analysis revealed that child challenging

behaviour was not the strongest predictor of negative or positive maternal measures, but positive child mood did emerge as a frequent predictor of both negative and positive parental outcomes.

The sixth chapter is a discussion of the studies contained in this thesis. It summarizes the findings and implications of the empirical research, and makes recommendations for future research and practice.

Chapter 2: Using Matched Groups to Explore Child Behaviour Problems and Maternal Well-Being in Children with Down Syndrome and Autism¹

¹ A version of this chapter has been accepted for publication as Griffith, G. M., Hastings, R. P., Nash, S., & Hill, C. (in press). Using Matched Groups to Explore Child Behaviour Problems and Maternal Well-Being in Children with Down Syndrome and Autism. *Journal of Autism and Developmental Disabilities*.

Abstract

Background. Although research on behaviour problems in children with autism and children with Down syndrome has led to generally consistent findings, existing studies suffer from considerable internal validity problems. In particular, researchers rarely use a matched group design.

Method. Mothers of children with Down syndrome, autism, and mixed aetiology intellectual disabilities, matched on child age, gender, and communication skills (n=19 in each group) completed measures of their child's adaptive and problem behaviours, their own parenting stress, and positive perceptions of their child.

Results. Children with autism were rated as having more problem behaviours and lower levels of social competence than children with Down syndrome and mixed aetiology intellectual disabilities. Mothers of children with autism scored lower on positive perceptions of their child, and higher on stress than the other two groups.

Conclusions. Contrary to previous research using unmatched groups, no statistically significant difference was found in levels of social competence or behaviour problems between children with Down syndrome and mixed aetiology intellectual disabilities. Our use of matched groups may have helped eliminate any confounding child variables that were not controlled for in previous studies. Further research could focus on using matched groups of children to control for potentially confounding variables.

Increasingly, researchers in the field of intellectual disabilities are interested in how the genetic or other diagnosis of an individual may predispose them to exhibit particular behaviour patterns, and the influence of these behavioural patterns on maternal outcomes, such as stress and depression (Abbeduto et al., 2004; Pisula, 2007). Behavioural profiles in genetic syndromes are part of what are known as 'behavioural phenotypes.' The phenotype of a syndrome reflects the increased likelihood of finding particular behavioural characteristics within affected individuals, for example, individuals affected by Angelman syndrome often display smiling and laughing behaviours (Oliver, Demetriades, & Hall, 2002). This probabilistic view of behavioural phenotypes suggests that genotype may predispose particular behaviours, but those behaviours are not inevitable (Dykens, 1995).

Although there is evidence for a strong genetic component underlying autism, it is not yet fully understood (Sykes & Lamb, 2007). Therefore, autism cannot be described as a genotype. However, there are strong behavioural features associated with autism, presumably related to underlying aetiology, which makes it a useful comparison group when exploring between-group differences. The focus of the present paper is on child behaviour and maternal outcomes within two diagnostic groups associated with a large proportion of cases of intellectual disability: autism and Down syndrome.

Children with autism are often reported as having more behaviour problems than children with mixed aetiology intellectual disabilities (Kasari & Sigman, 1997) and children with Down syndrome (Eisenhower, Baker, & Blacher, 2005; Sanders & Morgan 1997). In contrast, children with Down syndrome are reported as having fewer behaviour problems than other children with intellectual disabilities (Dykens

& Kasari, 1997; Ricci & Hodapp, 2003; Stores, Stores, Fellows, & Buckley, 1998). Recently, Blacher and McIntyre (2006) compared behaviour problems in children with autism, Down syndrome, and mixed aetiology intellectual disability and found that children with autism had the most problem behaviours, and children with Down syndrome the least. From the above studies on child behaviour problems, only two controlled for child variables (child gender and age) that may have been influential variables for child behaviour (Dykens & Kasari, 1997; Kasari & Sigman, 1997).

Although research on behaviour problems in children with autism and children with Down syndrome has led to generally consistent findings, existing studies suffer from considerable internal validity problems. In particular, researchers rarely use a matched group design. Therefore, the samples may differ on potentially significant variables including the child's age and gender. Researchers who have controlled child gender and age in making group comparisons do still tend to replicate the typical pattern of group differences (e.g., Dykens & Kasari, 1997). However, what is more problematic to internal validity is that children may vary considerably in their cognitive, language, and adaptive skills. This is perhaps most marked for children with autism, as some children have an intellectual disability and some do not. Therefore, it is often not clear whether intellectual disability or autism itself explains any observed group differences. In the current research, we explored differences in behaviour problems for children with intellectual disability who either had Down syndrome, autism, or other "mixed" diagnoses. Using a matching approach, we directly addressed threats to internal validity found in previous research.

In terms of parental outcomes, previous research findings mirror those found for group differences in child behaviour problems. Parents of children with autism

tend to report more stress and mental health problems than parents of children with mixed aetiology intellectual disability (Olsson & Hwang, 2001) and Down syndrome (Abbeduto et al., 2004; Holroyd & McArthur, 1976; Kasari and Sigman, 1997; Pisula, 2007; Sanders & Morgan, 1997). Parents of children with Down syndrome also report less stress than parents of children with mixed aetiology intellectual disability (Ricci & Hodapp, 2003; Roderigue, Morgan, & Geffken, 1992). Children with Down syndrome are more sociable than children with some other diagnoses (Kasari & Freeman, 2001; Pitcairn & Wishart, 1994), which may influence parental outcomes. However, no studies have directly examined whether increased sociability has an impact on parental well-being. This putative relatively positive outcome for parents of children with Down syndrome has been termed a “Down syndrome advantage” (Hodapp, Ly, Fidler & Ricci, 2003).

Research studies of psychological well-being in parents of children with autism and Down syndrome suffer from similar internal validity problems as the research focused on child behaviour problems. However, some researchers have adopted a matching procedure to reduce threats to internal validity in the exploration of the Down syndrome advantage (Cahill & Glidden, 1996; Stoneman, 2007). Stoneman (2007) found that both mothers and fathers of children with Down syndrome reported fewer symptoms of depression, and were also observed to engage in warmer parenting when compared with parents of children with mixed aetiology intellectual disabilities. However, when the variance due to familial income was removed, the group differences disappeared. Cahill and Glidden (1996) first compared an unmatched sample of children with Down syndrome and those with mixed aetiology intellectual disability, and found that parents of children with Down syndrome reported less stress. However, once the children were matched on

level of functioning, age, parental income, and parental marital status, this difference disappeared. It may be that child variables and family demographic differences underlie the Down syndrome advantage. Thus, one would predict that differences in parental stress outcomes for diagnostic groups may be accounted for by patterns of difference in the children's behaviour problems. Recent research data support this hypothesis (e.g., Abbeduto et al., 2004; Blacher & McIntyre; 2006).

The main aim of the present study was to adopt a close matching design to minimize threats to internal validity and to explore diagnostic group differences between autism and Down syndrome. First, all of the children in the sample had an intellectual disability (including all children with autism). Children were then matched on chronological age, gender, and also language/communication ability. We chose the latter variable because of recommendations to control for language skills when making any comparison between children with and without autism (Charman, 2004), and because children's language skills have also been found to be related to maternal outcomes (Most, Fidler, LaForce-Booth & Kelly, 2006). Using this matched design, we explored child behaviour and mental health problems in children with autism and Down Syndrome, and whether differences in child variables affected maternal well-being. Existing research has included a limited range of parental outcome measures and so we included a broad range of individual negative (stress, mental ill-health) and positive (positive perceptions, positive affect, and life satisfaction) adjustment outcomes, as well as dyadic and family outcomes (marital adjustment, family satisfaction). We hypothesized that any diagnostic group differences found for child behaviour would explain maternal outcome differences between the groups.

We also explored a further tentative hypothesis. There is some evidence to suggest that children with Down syndrome are more socially able than other children with intellectual disabilities (Kasari & Freeman, 2001; Pitcairn & Wishart, 1994). Thus, we explored whether this difference would emerge in our closely matched groups and whether such a difference might explain group patterns in maternal outcomes.

Method

Participants

After matching (see Procedure), there were 57 mothers with a child with intellectual disability who participated in this study: 19 children had Down syndrome, 19 had autism, and 19 had various other diagnoses. Of the 19 in the 'mixed' group, seven had cerebral palsy, three had epilepsy, one had Attention Deficit/ Hyperactivity Disorder, one had Pallister-Killian syndrome, and the remaining seven had an intellectual disability with unknown aetiology. The diagnoses were based on parental report, and we did not have access to clinical notes to establish the validity of these reports. Three children with Down syndrome also had a co-diagnosis of autism, but none of the children with mixed aetiology intellectual disabilities did so. Across the total sample, the children ranged in age from 4 years 3 months to 18 years old (mean = 10 years 4 months), and there were 18 girls and 39 boys (13 boys and 6 girls in each group). According to mothers' ratings on the Vineland Adaptive Behaviour Scales (VABS: Sparrow, Balla, & Cicchetti, 1984), two children were classified as having an 'adequate' level of adaptive skills (based on the VABS composite score). However, these were two children with autism, who were at the lowest end of the adequate range. Twenty one

children were classified as having a mild to moderate developmental delay, and 34 were classified as having a severe/profound developmental delay.

Demographic and child adaptive behaviour characteristics for the three groups are summarized in Table 2.1. The variables used for matching showed very similar mean levels and variance across the three groups. One-way ANOVA and chi-square tests (for dichotomous variables) were used to explore group differences on these and all other variables. No statistically significant group differences on child characteristics emerged. The only group difference was that mothers of children with Down syndrome of similar chronological age were older than the other mothers, and this difference was borderline statistically significant ($F(2, 53) = 3.15$, $p = .051$).

Table 2.1. Matched Group Characteristics- Child and Maternal Variables

Matching Variables	Child Aetiology		
	Autism	Down Syndrome	Mixed Aetiology
Child age in years – Range, Mean (SD)	4.3-17.3 years 10.16 (3.86)	5.3-17.4 years 9.98 (4.04)	4.4-18.0 years 9.84 (4.18)
Child VABS ¹ Communication standard score – Range, Mean (SD)	19-88 39.89 (16.51)	19-73 40.05 (14.18)	19-60 40.47 (12.25)
VABS Socialization standard score - Range, Mean (SD)	19-116 50.00 (22.73)	19-100 60.58 (17.79)	19-76 49.74 (15.26)
VABS Daily Living Skills standard score – Range, Mean (SD)	19-60 32.11(15.43)	19-78 40.89 (21.29)	19-98 34.32 (13.65)
VABS Adaptive Behaviour Composite – Range, Mean (SD)	19-79 37.63 (15.57)	20-63 43.89 (13.12)	19-71 38.26 (13.65)
Maternal age - Mean (SD)	38.63 (5.71)	43.74 (7.73)	38.89 (7.51)
Maternal educational level	University level or equivalent (%) 36.9	47.4	31.6
Marital status	Married or living with partner (%) 73.7	84.2	79

¹ Vineland Behavior Adaptive Scales

Measures

Demographic information was collected using a postal questionnaire (see Table 2.1 for variables, and Appendix 1). Mothers were interviewed over the telephone to complete the VABS.

Child Measures. The VABS (Sparrow et al., 1984) is a semi-structured interview, which was used to assess the adaptive skills of the child. The VABS assesses four domains: Socialization, Daily Living skills, Communication, and Motor Skills (used for children under seven years of age only), and an overall composite score can also be obtained.

Child behaviour problems were assessed using two measures, the Behaviour Problems Inventory (BPI: Rojahn, Matson, Lott, Ebensen, & Small, 2001, see Appendix 2), and the Reiss Scales for Children's Dual Diagnosis (Reiss & Valenti-Hein, 1990, see Appendix 3). The BPI has 52 items, which measure self-injurious behaviour (e.g., hitting head, self-scratching), stereotypical behaviours (e.g., twirling things, rocking back and forth), and aggressive/destructive behaviours (e.g., destroying things, biting others). Raters indicate the frequency of a particular behaviour on a five-point Likert scale ranging from 'never' to 'hourly'. The BPI frequency scores have been reported to have good reliability and construct validity (Rojahn et al., 2001). The Reiss Scales are a 60 item measure designed to assess psychopathology in children with intellectual disabilities (Reiss & Valenti-Hein, 1990). Raters score each item on a three point scale (No Problem, Problem, or Major Problem). There are 10 subscale scores (attention deficit, anger, anxiety, conduct disorder, depression, autism, psychosis, self-esteem, somatoform and withdrawn behaviours) as well as a total score. The Reiss scales also have good psychometric properties (Reiss & Valenti-Hein, 1994).

The social competence scale of the Nisonger Child Behaviour Rating Form (NCBRF: Aman, Tasse, Rojahn, & Hammer, 1996, see Appendix 4) was used to assess children's positive behaviour including calm/compliant behaviours (e.g., accepting redirection) and adaptive/social behaviours (e.g., shared with or helped others). The social competence scale includes ten items rated from "not true" to "completely or always true". The NCBRF has excellent psychometric properties (Aman et al., 1996), and Cronbach's alpha coefficient for the total social competence score was .87 in the present study.

Maternal Measures. General maternal stress related to having a child with disability in the family was measured using the Parent and Family problems subscale from the Questionnaire on Resources and Stress –short form (QRS-F; Friedrich, Greenburg, & Cmic, 1983, see Appendix 5). Five items were excluded from the subscale as they have been identified as a robust measure of depression (Glidden & Floyd, 1997) and we wished to reduce potential measurement overlap. The Kuder-Richardson coefficient for the present total sample was .91. The Positive Contributions Scale from the Kansas Inventory of Parental Perceptions (KIPP; Behr, Murphy, & Summers, 1992, see Appendix 6) was used to measure mothers' perceptions of the positive contributions the child has brought to them (such as happiness and fulfilment, learning patience, having a new perspective on life), to the wider family (e.g., bringing the family closer together), and the child themselves (e.g., is fun to be around). This scale has 50 items which are rated on a four point agreement scale. A total score was used, and this has strong reliability for parents of children with intellectual disabilities (Hastings, Beck, & Hill, 2005).

General maternal well-being was measured using two negative scales and two positive scales. On the negative side, maternal mental health was assessed using the

Hospital Anxiety and Depression Scales (Zigmond & Snaith, 1983, see Appendix 7) which includes seven anxiety and seven depression items and has been widely used in community samples of parents of children with disabilities, with excellent psychometric properties (e.g., Hastings et al., 2005). The Positive Affect Scale used in the current study was derived by extracting the ten positive affect items from the Positive and Negative affect scale (PANAS: Watson, Clark, & Tellegan, 1988, see Appendix 8). Mothers were asked to rate to what extent these ten items apply to them at the present moment on a Likert-type scale ranging from “very slight or not at all” to “extremely”. Cronbach’s alpha for the present sample was .89. Overall life satisfaction was measured using the Satisfaction With Life Scale (Diener, Emmons, Larsen, & Griffin, 1985, see Appendix 9). This is a five-item scale that asks participants to indicate their degree of agreement or disagreement to statements such as “In most ways, my life is close to ideal” on a seven-point Likert-type scale. Cronbach’s alpha coefficient was .85 for the present sample of mothers.

The final two maternal measures were focused on dyadic and family adjustment as opposed to individual well-being. The Golombok Rust Inventory of Marital State (Rust, Bennum, Crowe, & Golombok, 1990, see Appendix 10) assesses marital discord and overall marital satisfaction and was completed by mothers who were living with a partner. Respondents rate 28 items (e.g., “We both seem to like the same things”, “I no longer feel I can really trust my partner”) on a four-point Likert-type scale ranging from “Strongly Disagree” to “Strongly Agree”. A total score was used, which in the current study had a Cronbach’s alpha of .80. Higher scores indicate more dyadic adjustment problems. The Family Satisfaction Scale was used to measure family cohesion and adaptability (Olson & Wilson, 1982, see Appendix 11). This measure includes items such as “How satisfied are you with

how fair the criticism is in your family?” and “How satisfied are you with the amount of time you spend together as a family?” This 14 item measure uses a five-point Likert-type response scale, which ranges from “Dissatisfied” to “Extremely Satisfied”. Cronbach’s alpha for the present sample was .94.

Procedure

The participants in the present study were a sub-sample from a larger survey study of families of children with intellectual disabilities (Hastings et al., 2005). The families were recruited via their child’s school, information packs about the research project, including a response form and a business reply envelope were distributed throughout schools for children with intellectual disabilities. Once the response forms had been received, separate questionnaire packs and consent forms were posted to the primary caregiver, and when available, the secondary caregiver. A total of 139 mothers of children with various forms of intellectual disabilities participated in the larger project and completed postal questionnaires along with a telephone interview for the VABS.

The parent-reported diagnoses of the children in the full study were 26 with Down syndrome, 54 with autism, and 59 with other diagnoses associated with their intellectual disabilities. Given that Down syndrome was the smallest group, we attempted to match a child with autism and a child from the mixed diagnostic group to each child with Down syndrome. Three matching criteria were used: (a) child gender, (b) child age, and (c) the communication standard score from the VABS. All children were first matched on gender. For age, we attempted to match children born within 18 months of each other. This was not possible for six children with Down syndrome. However, the children in these cases were all over the age of eight years and given that the rate of development may be slower as children enter middle

childhood (Ramey & Ramey, 1998), this was deemed acceptable. In every case, no matched child was more than 28 months different in age to their matched child with Down syndrome. This matching procedure did have the effect of achieving well-matched groups.

The criterion for matching on communication score was that all three children's scores had to be within one standard deviation of each other. There were three cases where a match within a standard deviation could not be found, but these children were included due to a very close match on chronological age. Overall, we failed to find reasonable matches for seven of the participants with Down syndrome. This was predominantly due to a lack of females with autism available for matching, and missing VABS data for one child. The matching procedure led to very closely matched groups on the selected variables (see Participants and Table 2.1).

Results

Group Differences for Child Behaviour Measures

A series of one-way between-subjects ANOVAs were conducted on maternal ratings of their child's problem behaviours. Where a significant group effect was found, post-hoc Tukey's tests were used to explore pairwise differences. Mean scores for each group and the effect sizes for the pairwise comparisons are summarized in Table 2.2. There were eleven statistically significant group effects on child behaviour measures. There was a group effect on child social competence ($F(2, 54)=7.28, p=.002$), and frequencies of self-injurious ($F(2, 53)=7.95, p=.001$), and stereotypical behaviour ($F(2, 53)=6.07, p=.004$). Post-hoc analysis revealed that this was due to mothers rating their child with autism as having significantly lower social competence, and engaging in higher frequencies of self injurious and stereotypical behaviour than children with Down syndrome or mixed aetiology

intellectual disability. There were also significant group differences on the Reiss scales of anger ($F(2, 54)=5.29, p=.008$), anxiety ($F(2, 54)=12.38, p=.000$), depression ($F(2, 54)=3.89, p=.026$), autism ($F(2, 54)=10.30, p=.000$), psychosis ($F(2, 54)=9.78, p=.000$), self-esteem ($F(2, 54)=6.39, p=.003$), withdrawn behaviours ($F(2, 54)=9.48, p=.000$), and Reiss total score ($F(2, 54)=9.55, p=.000$).

Post-hoc analyses showed that mothers rated their children with autism as having significantly higher levels of anger, anxiety, depression, autism, psychosis, self-esteem, withdrawn behaviours, and the total Reiss score than mothers of children with Down syndrome. Mothers also rated their children with autism as being significantly higher on the Reiss subscales of anxiety, autism, psychosis, and also on the Reiss total score than mothers of children with mixed aetiology intellectual disabilities. There were no significant group differences between children with Down syndrome and those with mixed aetiology intellectual disabilities. All pairwise statistically significant effects were also associated with large effect sizes (Cohen, 1992).

These group analyses were repeated without the three individuals from the Down syndrome group with a co-diagnosis of autism and their matched counterparts to examine whether this may have influenced the results. The pattern of results found was the same, with the exception that an additional group difference was found on the aggressive/destructive behaviour subscale of the BPI ($F(2,50)=3.67, p=.033$). Post-hoc tests showed that mothers of children with autism rated their child as engaging in significantly higher frequencies of aggressive-destructive behaviour than mothers of children with Down syndrome.

Table 2.2. Group Means, Standard Deviations, and Effect Sizes (Cohen's *d*) for Child Behaviour Variables

Child Behaviour Measures	Means (SD)			Effect Sizes		
	Autism	Down Syndrome	Mixed ID	Autism vs. Down Syndrome	Autism vs. Mixed ID	Down syndrome vs. Mixed ID
Nisonger CBRF ¹ - social competence subscale	8.98 (5.41)	14.95 (4.25)	14.19 (5.95)	-1.22**	0.91**	0.15
BPI ² - Frequency of self-injurious behaviour	12.06 (8.61)	3.51 (4.42)	5.43 (6.83)	1.25***	0.85*	0.33
BPI - Frequency of stereotypical behaviour	36.42 (23.59)	17.22 (18.10)	16.75 (16.86)	0.91*	0.96**	0.01
BPI - Frequency of aggressive/destructive behaviours	11.89 (11.31)	5.91 (7.77)	6.16 (9.20)	0.62	0.55	-0.03
Reiss ³ - attention deficit	3.74 (2.44)	2.06 (2.69)	2.42 (2.19)	0.65	0.57	0.15
Reiss - anger	5.41 (2.74)	2.31 (2.78)	3.16 (3.52)	1.12**	0.71	-0.27
Reiss - anxiety	3.47 (2.73)	0.47 (0.84)	1.21 (1.75)	1.48***	0.98**	-0.54
Reiss - conduct disorder	2.21 (2.44)	0.89 (1.73)	1.05 (2.20)	0.62	0.50	-0.08
Reiss - depression	2.42 (1.98)	0.85 (1.22)	1.58 (1.89)	0.95*	0.43	-0.45
Reiss - autism	3.91 (2.45)	1.37 (1.95)	1.10 (1.85)	1.14***	1.29***	0.14
Reiss - psychosis	4.47 (2.06)	1.70 (1.76)	2.53 (2.11)	1.45***	0.93*	-0.43
Reiss - self-esteem	2.18 (1.85)	0.42 (0.84)	1.21 (1.68)	1.23**	0.55	-0.59
Reiss - somatoform	0.47 (0.84)	0.37 (0.95)	0.68 (1.00)	0.12	-0.23	-0.32
Reiss - withdrawn	5.31 (2.94)	1.84 (2.93)	2.00 (2.43)	1.18***	1.23*	-0.06
Reiss - total score	37.29 (16.77)	14.04 (15.50)	19.28 (19.14)	1.44***	1.00**	0.30

p<.05; **p<.01; ***p<.001

¹ Nisonger Child Behaviour Rating Form

² Behavioural Problems Inventory

³ Reiss Scales for Dual Diagnosis

Group Differences for Maternal Well-Being

Maternal well-being was explored using between-subjects ANOVA. All analyses were repeated including maternal age in separate ANCOVA analyses, but this did not change the pattern of results and thus ANOVA results only are reported. There were two statistically significant group differences, one was on the Parent and Family Problems scale ($F(2, 52)=14.26, p=.000$), post-hoc analysis showed that mothers of children with autism reported more stress on the scale than mothers of children with Down syndrome. Furthermore, there was a significant group difference on the Positive Contributions scale ($F(2, 53)=5.84, p=.005$). Post-hoc analysis revealed that mothers of children with Down syndrome had significantly higher positive contributions scores than mothers of children with autism. Mean group scores and effect sizes for pairwise comparisons are summarized in Table 2.3. Again, statistically significant group differences were associated with large effect sizes. The above analyses were also repeated without the three individuals with a co-diagnosis of Down syndrome and autism and their matched counterparts to check whether this influenced the results. Again, the pattern of results remained unchanged.

To examine the hypothesis relating to the influence of child behaviour problems on maternal outcomes, group differences in maternal stress and positive perceptions were explored in a series of ANCOVA models where salient child behaviour problems variables (those showing statistically significant group differences) were introduced as covariates one at a time.

Table 2.3. Group Means, Standard Deviations, and Effect Sizes (Cohen's *d*) for Maternal Variables

Maternal Variables	Aetiology			Effect Sizes		
	Autism Mean (SD)	Down Syndrome Mean (SD)	Mixed ID Mean (SD)	Autism vs. Down Syndrome	Autism vs. Mixed ID	Down Syndrome vs. Mixed ID
QRSF ¹ Family problems (excluding depression)	8.64 (3.68)	3.48 (2.87)	3.88 (3.26)	1.56**	1.37**	-0.13
HADS ² anxiety	10.00 (4.47)	8.16 (4.18)	8.89 (2.77)	0.42	0.30	-0.21
HADS depression	6.89 (4.09)	5.53 (4.03)	5.16 (2.79)	0.34	0.49	0.11
PCS ³ total score	125.92 (13.87)	141.69 (22.26)	135.35 (18.32)	-0.85*	-0.59	0.31
Satisfaction with life	18.42 (6.21)	22.74 (7.51)	19.00 (5.90)	-0.63	-0.09	0.55
Family satisfaction	32.21 (9.89)	38.48 (11.02)	37.42 (9.31)	-0.60	-0.54	0.10
Positive affect scale	30.00 (7.73)	35.58 (8.03)	31.47 (6.49)	-0.68	-0.21	0.53
GRIMS ⁴ total	30.15 (16.36)	30.99 (15.21)	33.51 (13.04)	-0.05	-0.23	-0.18

* $p < .05$ ** $p < .001$ ¹ Questionnaire on Resources and Stress –short form² Hospital Anxiety and Depression Scale³ Positive Contributions Scale⁴ Golombok Rust Inventory of Marital State

The group difference on mothers' perception of positive contributions was no longer statistically significant when either social competence ($F(2,54)=3.50$, $p=.30$) or behaviour problems were controlled for (self-injurious behaviour, $F(2,53)=7.95$, $p=.17$, stereotyped behaviour, $F(2,53)=6.07$, $p=.10$, Reiss scales total score, $F(2,54)=9.55$, $p=.25$). However, the group difference on maternal stress remained after controlling for either social competence or any measure of behaviour problems.

To explore the possibility that maternal psychopathology might influence mother's ratings of their child's behaviour, an ANCOVA was run on child behaviour measures (Nisonger CBCL, BPI, and Reiss total scores) using maternal depression as a co-variate. All group differences in child behaviour problems reported earlier remained after controlling for maternal self-reported symptoms of depression.

Exploration of Effect sizes

The mean Vineland socialization score was higher in children with Down syndrome than in the other two groups (see Table 2.1). To further explore this finding, the socialization scores were compared using ANOVA and there was no evidence of a significant group difference ($F(2,54)=2.05$, $p=.14$). However, the effect sizes of the difference between children with Down syndrome and those with autism (Cohen's $d = .52$) and those with mixed aetiology intellectual disability (Cohen's $d = .65$) were in the moderate range. Thus, there is some support for the presence of a relative advantage in social behaviour for children with Down syndrome despite matching for age and gender. However, when we repeated the group comparisons for maternal stress and maternal positive perceptions introducing

VABS socialization scores as a covariate, these group differences still remained statistically significant.

Further moderate effects were found that are worthy of comment. Mothers of children with Down syndrome rated their children as having lower anxiety (Cohen's $d = .54$) and depression (Cohen's $d = .45$) on the Reiss scales than mothers of children with mixed aetiology intellectual disabilities. In terms of maternal outcomes, mothers of children with Down syndrome reported greater life satisfaction (Cohen's $d = .55$) and positive affect (Cohen's $d = .53$) than mothers of children with mixed aetiology intellectual disabilities.

Discussion

This is the first study of which we are aware in which child characteristics have been closely matched to investigate both social competence and problem behaviours in Down syndrome and autism, and to have explored relationships with maternal well-being once behaviour differences are controlled. We first examined the evidence for problem behaviour or social competence relating to child diagnosis. Mothers of children with autism rated their child as having significantly lower social competence, as well as engaging in higher frequencies of problem behaviour when compared to mothers of children with Down syndrome and mixed aetiology intellectual disabilities. This result is consistent with previous research where group matching has not been used to reduce internal validity threats (Eisenhower et al., 2005; Ricci & Hodapp, 2003; Stores et al., 1998). Contrary to previous research using unmatched groups, no statistically significant difference was found in levels of social competence or behaviour problems between children with Down syndrome and mixed aetiology intellectual disabilities. Our use of matched groups may have

helped eliminate any confounding child variables that were not controlled for in previous studies.

Turning to maternal outcomes, no significant differences were found in levels of anxiety, depression, marital satisfaction, positive affect, overall life satisfaction, and family satisfaction across the diagnostic groups. Reported well-being for mothers of children with autism was found to significantly differ from the other two groups on just two measures. Mothers of children with autism scored significantly higher on maternal stress than mothers of children with Down syndrome, and significantly lower on positive perceptions than both mothers of children with Down syndrome and mixed aetiology intellectual disabilities.

Although the difference for maternal stress remained even after controlling for child behaviour problems and positive social behaviour, the group differences for positive contributions did not. Thus, using a matched groups design, very few differences between groups were observed despite a broad range of measurement of maternal well-being. Therefore there was little evidence of a Down syndrome advantage (Hodapp et al., 2003). This is similar to previous research that controlled for child or socio-economic variables (Cahill & Glidden, 1996; Stoneman, 2007). However it is important to note that some moderate effect sizes were evident which are more consistent with the Down syndrome advantage. Mothers of children with Down syndrome rated their children as having lower anxiety and depression than mothers of children with mixed aetiology intellectual disabilities. Mothers of children with Down syndrome also reported greater life satisfaction and positive affect than mothers of children with mixed aetiology intellectual disabilities. Furthermore, mothers of children with autism scored lower on life satisfaction (Cohen's $d = .63$), family satisfaction (Cohen's $d = .60$), and on positive affect (Cohen's $d = .68$)

compared with mothers of children with Down syndrome. Research using larger matched groups is warranted to help further explore the issue.

Children with Down syndrome were rated higher on the VABS socialization domain than the other two groups (see Table 1). However, controlling for socialization scores did not affect the diagnostic group differences on maternal well-being. Thus, better child social skills did not seem to explain better maternal adjustment in the Down syndrome group. These trends are worthy of exploration using larger matched groups in future research, as Type II error may confound such findings within this small sample size.

The observed group differences for maternal well-being were unaffected by maternal age differences between the groups. It has been suggested previously that because the average age of mothers with children with Down syndrome is higher than among the general population (Olsen, Cross, Gensburg & Hughes, 1996), they may have better financial resources and greater life experience which may help to buffer the effects of having a child with a disability (Hodapp et al., 2003). Our data did not support this hypothesis, although 19 participants in each diagnostic group may be too small a sample for any differences to emerge. In terms of mothers of children with autism, although the child's behaviour problems do not seem to explain maternal stress differences there may be another variable associated with autism which does. For example, unlike in Down syndrome where there is no evidence of maternal personality traits associated with the behaviour patterns seen in the child, parents of children with autism may display aspects of the autism phenotype themselves, such as weaker central coherence and social rigidity (Bailey, Palferman, Heavey, & Le Couteur, 1998; Happe, Briskman, & Frith, 2001). These difficulties may place parents at risk for psychological problems themselves, and

could bias parental reports of child problem behaviours. Such processes could conceivably also apply to other genetic disorders associated with intellectual disability such as fragile X syndrome (Hessl, Dyer-Friedman, Glaser, Wisebeck, Barajas et al., 2001). A putative interaction between environmental risks (i.e., child behavioural difficulties) and parental genetic vulnerability that is linked to underlying shared characteristics with their child may be a potentially important area of further research. Parental adjustment may be related to the child's behaviour, their own genetic vulnerabilities, and/or an interaction between these risks. Autism is perhaps the clearest example of these putative effects, but further exploration in other genetic syndromes may also be warranted.

These conclusions need to be tempered with reference to some methodological points. First, our matched groups were rather small and power to detect even moderate sizes of group differences was therefore limited. Although the use of matched groups has added a degree of control and improved internal validity, this approach is likely to sacrifice any representativeness of the samples and thus external validity. Diagnosis was based on maternal report, and we do not have information on the aetiology of 7/19 children in the mixed aetiology intellectual disability group, these factors should be borne in mind as it may explain the differences to previous studies which used an mixed aetiology intellectual disability group (Ricci & Hodapp, 2003; Roderigue et al., 1992). Additionally, future research should make efforts to get clinical confirmation of the child's diagnosis.

The wide age range of the children should also be considered, as there may be variance in maternal outcomes attributable to the age of the child, this could be controlled for in future research by ensuring the children are from narrower age bands. Examination of the effect sizes in Tables 2.2 and 2.3 suggests some sizeable

group differences that are potentially clinically meaningful and might emerge as statistically significant with larger samples. Thus, studies with large matched groups are needed in future. The small sample sizes created via the matching approach also limited us to between-group analysis. With larger groups, it would be possible to begin to explore questions such as whether the strength of associations between child behaviour and parental adjustment differ between diagnostic groups. These differences are worthy of exploration in future research.

A second methodological point relates to our choice of maternal well-being measures. Although we chose a broader range of measures than many similar studies, these were all focused on aspects of maternal well-being and mothers' perceptions of their family relationships. We did not explore the putative impact of the child's disability on mothers' work life, broader social life, or even their day-to-day caring responsibilities. These variables might well influence maternal well-being in these and other domains if investigated in future research. Thirdly, the focus of this study was on mothers. In the future it is important to also explore effects on fathers, siblings, and other family members.

A final issue is that there are well-established links between socioeconomic position and both child disability and parental well-being (Emerson, Graham, & Hatton, 2006). Of relevance to the present research, Stoneman (2007) found that an apparent Down syndrome advantage was explained primarily by familial income differences. Emerson, Hatton, Llewellyn, Blacher, and Graham (2006) found that lower psychological well-being in mothers of children with intellectual disabilities when compared to mothers who did not have children with intellectual disabilities was explained to a large extent by differences in deprivation between the groups. We had only a poor proxy variable for socioeconomic position available in this

study (maternal education), which did not differ between the groups. Therefore, further research is needed to examine the influence of socioeconomic position on the well-being of parents of children with intellectual disability.

Research focusing on how different child diagnoses influence the behaviour of children, and in turn their parents, has significant implications for intervention strategies. For example, clarity about behaviour patterns that are likely to develop within a diagnostic group can lead to early detection and targeted early interventions to help reduce the future occurrence or severity of problematic behaviours. An understanding of the vulnerabilities in parental adjustment is also important. Our results suggest, for example, that parents of children with autism may be at increased risk for distress and could benefit from targeted psychological support (cf. Singer, Ethridge & Aldana, 2007). A combination of risks may also be salient for clinical services to monitor. Looking out for the emergence of damaging behaviours in certain diagnostic groups where parents are already experiencing considerable stress might be important in targeting scarce support resources. It is also possible that parental support offered by clinical services is best adjusted to take account of diagnostic group differences. However, this is an empirical question as yet very rarely even considered as a moderator variable in intervention outcome research.

Chapter 3. Psychological distress and well-being in mothers and fathers of children with Angelman, Cornelia de Lange, and Cri du Chat syndromes¹

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Abstract

Background. The current study focuses on mothers and fathers of children with three rare genetic syndromes that are relatively unexplored in terms of family experience; Angelman syndrome (AS) Cornelia de Lange syndrome (CdLS) and Cri du Chat syndrome (CdCS).

Method. Parents of children with AS (n=15), CdLS (n=16), CdCS (n=18), and a matched comparison group of parents of children with autism (n=20) completed questionnaires on both psychological distress (stress, anxiety, depression) and positive psychological functioning.

Results. Parents of children with AS consistently reported the highest levels of psychological distress, and parents of children with CdLS the lowest, with parents of children with CdC and autism scoring between these two. Positive psychological functioning were similar across the four aetiology groups.

Conclusions. Parents of children with rare genetic syndromes are at risk for high levels of stress and mental health problems. Methodological issues and the practical applications of these results are discussed.

Advancements in genetics research have led to a growing interest in the behavioural phenotypes associated with rare intellectual disability (ID) syndromes (Hodapp & Dykens, 2001). However, the families of children with rare genetic syndromes have been the focus of surprisingly few research studies. Most family research in this area has either ignored the aetiology of the child's ID or has focused on parents of children with more common conditions associated with intellectual disability, such as autism and Down syndrome (e.g., Hodapp, 1997; Olsson & Hwang, 2001; Sanders & Morgan, 1997; Stoneman, 2007). In the current study, the focus is on three rare genetic syndromes associated with characteristic behavioural phenotypes: Angelman, Cornelia de Lange, and Cri du Chat syndromes.

These three syndromes are of interest partly because they share behavioural features, including severe ID and the presence of behaviour problems, which have previously been associated with increased parental stress and mental health problems (Baxter, Cummings, & Yiolitis, 2000; Hastings et al., 2005b; Kasari & Sigman, 1997; Most, Fidler, Laforce-Booth, & Kelly, 2006). In a review of the literature we identified only five studies focusing on the families of children with these syndromes: three on parents of children with Cornelia de Lange syndrome, and one each on parents of children with Cri du Chat and Angelman syndrome. These studies were concerned with determining levels of parental stress or mental health problems, and examining whether child characteristics (e.g., behaviour problems, adaptive behaviour, and age) affect parental stress levels. The gender of the caregivers was not reported in four of these studies (although the vast majority of participants were described as mothers). Therefore, the term 'parents' will be used when the gender is unknown.

Parents of children with Cornelia de Lange syndrome (n=27) reported higher levels of child related parenting stress than parents of typically developing children, and high parental stress levels were related to lower child adaptability, severe ID and increased child age (Sarimski, 1997). Although Sarimski (1997) did not examine associations between child behaviour problems and parental well-being, Wulffaert et al. (2009) found that child behaviour problems were the strongest predictor of parental stress among 37 parents of children with Cornelia de Lange syndrome. Additionally, over one third of parents reached cut-off for “very high stress” on the Parenting Stress Index (PSI: Abidin, 1990). Richman, Belmont, Kim, Slavin, and Hayner (in press) focused on child behaviour problems and parental stress in children and young adults with Cornelia de Lange syndrome (n=25) and Down syndrome (n=23). Parental stress was significantly higher in parents of children with Cornelia de Lange syndrome, and 40% of parents scored above the 95th percentile for total stress scores on the PSI. Parental stress was associated with high levels of child self-injury, stereotypy, and lower levels of child pro-social and adaptive behaviour.

Hodapp, Wijma, and Masino (1997) recruited 99 parents of children with Cri du Chat syndrome. They found that parental stress levels were higher than reported by parents of children with mixed aetiology ID, and the strongest predictor of parental stress was child behaviour problems. Lower child adaptive behaviour was also a moderate predictor of increased parental stress. In the only study on families of children with Angelman syndrome (n = 22), van den Borne et al. (1999) examined both mothers (n=22) and fathers (n=15), and compared them to parents of children with Prader-Willi syndrome. The authors did not examine child behaviour problems, but focused on parental depression, self-esteem, and coping strategies. There were

no differences found between mothers and fathers, but some differences emerged between the two syndrome groups. Parents of children with Angelman syndrome reported higher self-esteem, but more loss of control (e.g., feeling “tied down” because of their child) than parents of children with Prader-Willi syndrome. Parental depression levels were fairly high for both groups of parents.

The present study was designed to develop research on the families of children with Angelman, Cornelia de Lange, and Cri du Chat syndromes to further understand the levels of stress, anxiety, and depression experienced by parents, and to explore any positive outcomes experienced by parents. In doing so, we also address five methodological issues: (1) Variability of child behaviour problems as a confounding factor when examining parental measures, (2) Mother-father differences, (3) Stresses associated with the rareness of the syndrome, (4) Positive as well as negative parental outcomes, and (5) The use of parents of children with autism as a ‘benchmark’ for parental distress among parents of rarer syndromes. Each of these issues is discussed briefly below.

The first methodological issue relates to the three studies of AS, CdLS and CdC which explored child behaviour problems and the association with parental stress (Hodapp et al., 1997; Richman et al., in press; Wulffaert et al., 2009). All three studies found statistically significant associations. However, it is not known whether the samples in these studies also included children who did not show any behaviour problems, as having behaviour problems was not an explicit inclusion criterion. Therefore, it is difficult to evaluate whether any family outcome differences between syndromes are influenced by large variations in behaviour problems within a group. In the present study, we recruited only families of children with one of the three rare syndromes who also had significant behaviour problems.

The second methodological issue is the importance of distinguishing the experiences of mothers from those of fathers. Most family research on rare genetic syndromes has generally focused on mothers, probably because they are often the primary caregiver when a child has a disability (Simmerman, Blacher, & Baker, 2001). Although evidence is equivocal as to whether mothers and fathers react differently to raising a child with an intellectual disability (e.g., McCarthy, Cuskelly, van Kraayenoord, & Cohen, 2006; Shin, Nahn, Crittenden, Flory, & Ladinsky, 2006; van den Borne et al., 1999), it is important to include fathers in family research not least because different parts of the family system are theoretically likely to be affected differently (MacDonald, Hastings & Fitzsimons, in press).

To what extent factors pertaining uniquely to the *rareness* of the child's syndrome affect family experiences is the third methodological issue. This is a question seldom explored within the family literature on rare genetic syndromes. Where associations with rarity have been identified, researchers have focused on characteristic behaviours of individuals with the syndrome (e.g., unusual facial movements in Rett syndrome) and how these might relate to parental stress (Hodapp, Dykens, & Masino, 1997; Laurvick et al., 2006). Other more general potential stressors that may be associated with having a child with a rare syndrome (e.g., more frequent medical complaints and procedures, difficulty in finding practitioners with any knowledge of the syndrome) have tended to be neglected in previous research.

The fourth methodological issue concerns growing interest in the putative positive impact of having a child with an intellectual disability. Existing data and theory suggest that the positive impact of the child on family members occurs concurrently with, and is independent of, any negative impact (e.g., Blacher & Baker, 2006; Hastings & Taunt, 2002). None of the existing studies on the families

of children with Angelman, Cornelia de Lange, or Cri du Chat syndromes have explored positive as well as negative psychological well-being.

Finally, there is a difficulty in choosing appropriate control groups for assessing the relative degree of parental negative or positive outcomes in rare genetic syndromes. Comparison groups in existing intellectual disability genetic syndrome research have included parents of typically developing children, and parents of children with other specific aetiologies (including relatively more common conditions such as Down syndrome). In the present study we selected families of children with autism and an intellectual disability as an appropriate comparison group. This decision was based on the grounds that parents of children with autism reliably report more psychological distress than parents of typically developing children, parents of children with an intellectual disability or developmental delay, parents of children with specific developmental conditions (Down syndrome, fragile X syndrome, Cerebral palsy), and parents of children with physical or mental health problems (Abbeduto et al., 2004; Blacher & McIntyre, 2006; Duarte, Bordin, Yazigi, & Mooney, 2005; Herring et al., 2006; Lewis et al., 2006; Mugno, Ruta, D'Arrigo, & Mazzone, 2007; Schieve, Blumberg, Rice, Visser, & Boyle, 2007; Rutgers et al., 2007). Therefore, if a given parental group scores similar to or higher than parents of children with autism, it is likely that the parental group in question is undergoing substantial stress.

The principal aims of the present study were to address the five methodological issues identified above, by comparing positive and negative well-being of mothers and fathers of children and adolescents with Angelman, Cornelia de Lange, and Cri du Chat syndromes, who display behaviour problems on at least a daily basis. Using an existing database, we also included a matched comparison

group of parents of children with autism and an intellectual disability to help assess the relative extent of psychological distress of parents of children with rare syndromes. Finally, we developed a measurement tool to explore the rare syndrome-related stressors experienced by parents in the syndrome groups only.

Method

Participants

In total, 69 families participated in the current study, 15 families of a child with Angelman syndrome (14 mothers, 12 fathers), 16 families of a child with Cornelia de Lange syndrome (15 mothers, 14 fathers), and 18 families of a child with Cri du Chat syndrome (18 mothers, 13 fathers). The matched autism comparison group consisted of 20 families of children with autism and an intellectual disability (20 mothers and 7 fathers). Data for this group were taken from an earlier study on families of children with ID (Hastings, Beck, & Hill, 2005). Demographic details for all four aetiology groups are summarised in Table 3.1. All parents were the biological parents of their child, except for five of the children with Cornelia de Lange syndrome (four were adopted, and one was fostered).

A series of one-way between-subjects ANOVAs and chi-square tests were conducted on demographic variables across the four groups. Significant group effects were found on maternal age ($F(3,63)=6.39, p=.001$), and on the Vineland Adaptive Behavior Scales (VABS; Sparrow, Balla, & Cicchetti, 1984; Sparrow, Cicchetti, & Balla, 2005) adaptive behaviour composite ($F(3,65)=11.41, p=.000$). Post-hoc tests were then used to explore pairwise differences and these are displayed in Table 3.1.

Table 3.1. Demographic information on the four aetiology groups, with post-hoc analysis.

Demographics	Angelman syndrome (n=15)	Cornelia de Lange syndrome (n=16)	Cri du Chat syndrome (n=18)	Autism (n=20)	Post hoc test
Child age range	3.0-18.7 years	5.0-18.6 years	2.2-16.0 years	3.7-15.2 years	--
Child age (Mean, SD)	10.07 (4.79)	11.75 (3.49)	7.83 (4.66)	9.30 (3.37)	--
Child gender (n males)	10 males	10 males	4 males	16 males	
VABS composite score	41.13 (10.68)	39.44 (10.69)	53.44 (7.18)	38.00 (7.16)	CdC>CdLS**, AS**, Autism**
Maternal age, range and mean (SD)	32-50 years 41.79 (6.04)	37-65 years 47.31 (8.90)	31-50 years 39.56 (5.22)	27-47 years 37.89 (6.18)	CdLS >CdC**, Autism**
Paternal age, range and mean (SD)	32-48 years 42.38 (4.82)	30-65 years 47.60 (10.38)	31-48 years 41.92 (4.92)	28-47 years 39.14 (6.41)	--
% of primary caregivers married or living with a partner	87.6%	93.6%	66.6%	70%	--
% of families earning below £25,000	28.6%	33.3%	47.1%	75%	--

*p<.05

**p<.01

Of particular note is that the sample of children with Cri du Chat syndrome had significantly better overall adaptive behaviour than children with Angelman syndrome, Cornelia de Lange syndrome, and autism, although all children were classified as having a low level of functioning (<70 on the composite score).

Measures

The VABS (Sparrow et al., 1984) was used to interview mothers of children with autism and ID over the telephone. The VABS is a semi-structured interview, used to assess the adaptive skills of the child. The VABS assesses four domains: Socialization, Daily Living skills, Communication, and Motor Skills (used for children under seven years of age only), and an overall adaptive behaviour composite score is obtained by combining the scores of the four domains. For the three rare syndrome groups, the primary caregivers were interviewed over the telephone using the VABS- Second edition (Sparrow et al., 2005), which measures the same four domains as the earlier version of the VABS. The dataset with the parents of children with autism was collected prior to the publication of the VABS-II, hence different versions of the VABS were used. The VABS-II has good test-retest reliability, with correlations ranging from .80 to .95, and inter-rater reliability, with correlation coefficients from .75 to .85 (Sparrow et al., 2005).

In addition, parents of children in the three rare syndrome groups completed five questionnaire scales. The Parent and Family problems subscale from the Questionnaire on Resources and Stress – short form (QRS-F: Friedrich, Greenburg, & Crnic, 1983, see Appendix 5) was used to measure general parental stress related to having a child with a disability. Five items were excluded from the original subscale as they have been identified as a robust measure of depression and we

wished to reduce potential measurement overlap (Glidden & Floyd, 1997). Parents were asked to circle either “True” or “False” on 15 items (e.g., “Other members of the family have to do without things because of N”, and “N is able to fit into the family social group”). The Kuder-Richardson coefficient (equivalent to Cronbach’s alpha for scales with dichotomous items) for mothers of children with rare syndromes in the present research was .78, and for fathers .89.

The Hospital Anxiety and Depression Scales were used to assess parental mental health (Zigmond & Snaith, 1983, see Appendix 7). Although originally developed for residential populations, this measure has been used extensively in community research. Research with various populations has also suggested that the HADS has good agreement with other mental health measures such as the Center for Epidemiological Studies Depression scale (e.g., Katz, Kopek, Waldron, Devin, & Tomlinson, 2004). The HADS contains 14 four point items, with seven assessing depression (e.g., “I feel as if I am slowed down”) and seven assessing anxiety (e.g., “I get sudden feelings of panic”). The HADS has been widely used in community samples of parents of children with ID, and has excellent psychometric properties (e.g., Hastings, Beck, & Hill., 2005a). Cronbach’s alpha for the present sample of mothers of children with rare genetic syndromes was .88, and for fathers .91.

The Positive Affect Scale was derived by extracting the ten positive affect items from the Positive and Negative affect scale (PANAS: Watson, Clark, & Tellegan, 1988, see Appendix 8). Parents were asked to rate to what extent the ten items such as “strong” and “interested” have applied to them in the past week, on a Likert-type scale ranging from “very slight or not at all” to “extremely.” Internal consistency within the current sample was good with a Cronbach’s alpha score of .91 and .92 for mothers and fathers respectively.

The Positive Gain Scale (MacDonald et al., in press; Pit-ten Cate, 2003, see Appendix 12) assesses the direct positive aspects of having a child with a disability. Seven items including “Since having this child I feel I have grown as a person” and “Since having this child, my family has become closer to one another” are rated using a 5-point Likert scale from “0=strongly agree” to “4=strongly disagree”. The lower the score, the higher the positive gains reported by parents. Cronbach’s alpha for the present sample of mothers and fathers was .71 and .75, respectively.

The final questionnaire measure was devised for the purposes of the present research. The Genetic Syndrome Stressors Scale (GSSS, see Appendix 13) was designed to assess parental stressors relating to rare genetic disorders. Two sources of information were used to generate items for the GSSS. First, existing measures of difficulties associated with the parenting of children with an intellectual disability were reviewed. Second, semi-structured telephone interviews were conducted with six parents of children with Angelman, Cri du Chat, or Cornelia de Lange syndromes. These parents were asked to describe the stressful aspects of caring for their child, especially stressors that might be more likely to be present for families of children with rare syndromes. The resulting questionnaire had 14 items. Based on a total score across all 14 items, Cronbach’s alpha for the current sample was .83 and .87 for mothers and fathers respectively. Preliminary exploration of the concurrent validity of the GSSS showed that it is moderately positively associated with maternal anxiety (Pearson’s $r = .59$), depression ($r = .55$), and stress ($r = .61$), and negatively correlated with Positive Affect ($r = -.41$). For fathers, the GSSS was positively correlated with anxiety ($r = .47$) depression ($r = .52$), and stress ($r = .46$) and negatively correlated with Positive Affect ($r = -.33$). These data suggest that the GSSS has good face validity, internal consistency, and concurrent validity.

Procedure

This study was part of a wider project concerned with the behavioural functioning of children with the three rare syndromes as well as family adjustment. Sixty families (20 from each rare syndrome group) were recruited for the wider study with. All children: (1). Had a clinical or genetically confirmed diagnosis of either Angelman syndrome, Cornelia de Lange syndrome, or Cri du Chat syndrome; (2). Were between 2 and 19 years of age at the time of the study, and (3). Displayed self-injurious or other aggressive behaviour on at least a daily basis.

The majority of the 60 families ($n = 48$) were recruited from a database held by the research team. All families on this database were mailed a letter and an information leaflet explaining the nature of the research and the inclusion criteria (See Appendix 14). A researcher made telephone contact within seven days of mailing the information to determine whether potential participants met the three inclusion criteria for the study. The Challenging Behaviour Questionnaire (CBQ; Hyman, Oliver, & Hall, 2002, see Appendix 15) was used to determine the frequency of child aggressive or self-injurious behaviour. If the child had a confirmed clinical diagnosis of one of the three syndromes, was in the required age range, and was reported to engage in these problem behaviours at least once per day, they were included in the current study.

Of the 118 families initially screened in this manner, 62 (53%) met inclusion criteria for the study and 48 consented to take part. The remaining 12 families were recruited through mailing flyers (see Appendix 16) to families via national parent syndrome support groups, flyers being posted on the syndrome support group's websites, in newsletters and announcements at family conferences. Participants recruited in this manner were screened using the same procedure as above. Following

screening, parents were mailed a consent form, a detailed information sheet about the wider study, and a demographic questionnaire pack (see Appendix 17). Once consent was received, the family questionnaire packs were mailed, and the VABS-II was conducted via the telephone with the main caregiver within two weeks of the questionnaire pack being sent. Families were followed up by telephone if the questionnaire packs had not been returned within four weeks of mailing.

Of the 60 families recruited for the wider study, parents from 49 families completed the parental questionnaires (47 mothers and 38 fathers). The missing data were due to: two families being pilot participants for the main research study who were not asked to complete the questionnaire pack, and nine families not responding to requests to complete the questionnaire pack despite reminders. When only one parent completed a questionnaire pack, this was due either to divorce or separation (missing data from seven fathers and two mothers) or because they did not respond to requests to complete the questionnaire pack (three fathers).

The data from parents with a child with autism (autism diagnosis was based on parental report of received diagnosis) in the present study were taken from a larger study of families of children with an intellectual disability (Hastings et al., 2005a). The children with autism had to meet two additional inclusion criteria to be included in the research. First, the children had to display either aggressive or self-injurious behaviour on at least a daily basis. This was determined using the Behavior Problems Inventory (BPI: Rojhan, Matson, Lott, Esbenson, & Small, 2001, see Appendix 3). Children who were rated as engaging in any aggressive or self-injurious behaviour either daily or hourly on the frequency scale of the BPI were eligible for inclusion. Second, children had a VABS adaptive behaviour composite score of <70. This process resulted in the selection of 20 families whose child with

autism met both criteria. The parents of the children with autism and an intellectual disability completed the HADS, the QRS-F Parent and Family problems subscale, and the Positive Affect scale via postal questionnaire. The Positive Gain Scale and the GSSS were not used with the autism group.

Results

Clinical levels of anxiety and depression

Using a cut-off score of 11 on both the anxiety or depression scale of the HADS, as recommended by Zigmond and Snaith (1983), a higher percentage of parents in this study had likely clinical levels of symptoms compared with normative UK data (Crawford, Henry, Crombie, & Taylor, 2001: see Table 3.2). In particular, a much higher percentage (71.4%) of mothers of children with Angelman syndrome was at or above clinical cut-off for anxiety than the other three aetiology groups (range 33.3%-55%). Due to the small sample size, the assumptions for chi-square tests were not met and so one sample binomial tests were used to determine whether more mothers and fathers of the three rare syndromes and autism reported clinical levels of anxiety and depression than in the normative population (Crawford et al., 2001). The observed distributions differed significantly from the normative distribution for both mothers and fathers on anxiety and depression ($p < .05$) in all but one of the four aetiology groups. The one exception was fathers of children with Cornelia de Lange syndrome, who did not differ significantly from the male normative population on either anxiety or depression.

Table 3.2. The number of mothers and fathers at or above clinical cut off levels for anxiety and depression.

	Number of mothers/females reaching clinical cut off		Number of fathers/males reaching clinical cut off	
	Anxiety	Depression	Anxiety	Depression
Angelman Syndrome	10/14 ¹ (71.4%)	3/14 (21.4%)	5/12 (41.6%)	4/12 (33.3%)
Cri du Chat Syndrome	7/18 (38.9%)	4/18 (16.7%)	4/13 (30.8%)	2/13 (15.4%)
Cornelia de Lange Syndrome	5/15 (33.3%)	5/15 (33.3%)	2/14 (14.3%)	0/14 (0%)
Autism and ID	11/20 (55%)	3/20 (15%)	2/6 (33.3%)	0/6 (0%)
Normative population ²	12%	4%	6%	2%

One sample binomial tests were again used to determine whether the likelihood of meeting clinical cut off differed between syndrome groups. For mothers, it was found that the likelihood of reporting clinical levels of anxiety was significantly greater for mothers of children with Angelman syndrome than mothers of children with Cri du Chat ($p=.004$) and Cornelia de Lange syndrome ($p=.002$). Mothers of children with autism were significantly more likely to report clinical levels of anxiety than mothers of children with Cornelia de Lange syndrome ($p=.037$). There were no significant differences between syndrome groups on the likelihood of mothers meeting the clinical cut-off for depression.

For fathers, the likelihood of reporting clinical levels of anxiety was significantly greater for fathers of children with Angelman syndrome than fathers of children with Cornelia de Lange syndrome ($p=.031$). The likelihood of reporting

¹ These figures indicate how many out of the total number of mothers or fathers reach clinical cut off scores.

² Normative scores based on Crawford et al. (2001).

clinical levels of depression was significantly greater for fathers of children with Angelman syndrome than for fathers of children with autism ($p=.001$) and Cornelia de Lange syndrome ($p=.000$). Fathers of children with Cri du Chat syndrome were also more likely to report clinical levels of depression than fathers of children with autism ($p=.000$) and Cornelia de Lange syndrome ($p=.000$).

Group differences on maternal and paternal measures

Between-group ANOVAs were used to explore maternal and paternal data across all four aetiology groups. Group differences were found on maternal ($F(3,62)=5.61, p=.002$), and paternal ($F(3,41)=6.34, p=.001$) ratings of parental stress. When a significant group effect was found, post-hoc Tukey's tests were used to examine pair-wise differences, and these are summarised in Table 3.3. Where there were statistically significant group effects, the analyses were repeated including maternal age and child adaptive behaviour scores as covariates in separate ANCOVAs. These analyses did not change the pattern of results, and thus ANOVA results only are reported here. Additionally, all analyses were repeated using non-parametric tests (Kruskell-Wallis) due to the likelihood that the variables would not be normally distributed in these relatively small samples. The analyses again confirmed the results from the ANOVAs. The general pattern of results revealed that mothers of children with Angelman syndrome reported the highest scores on negative outcomes, mothers of children with Cornelia de Lange syndrome the lowest, with mothers of children with Cri du Chat syndrome and autism being in between. There was only one statistically significant group effect for maternal stress ($F(3, 61) = 5.61, p=.002$), and post-hoc analysis showed that this was related to mothers of children with Angelman syndrome reporting significantly higher stress levels than mothers of

children with Cornelia de Lange syndrome, Cri du Chat syndrome, and autism. The positive impact of the child on the family and maternal positive affect did not differ significantly between groups.

The paternal measures showed a similar pattern to those for mothers, although the mean scores for fathers were lower. Fathers of children with Angelman syndrome reported the highest scores for negative outcomes, fathers of children with Cornelia de Lange syndrome the lowest, with fathers of children with Cri du Chat syndrome and autism being in between. There was only one statistically significant group effects for paternal stress ($F(3, 41) = 6.34, p=.001$), and post-hoc analysis showed that this was related to fathers of children with Angelman syndrome reporting significantly higher levels than parents of children with Cornelia de Lange syndrome. and there was no significant group effect when examining positive outcomes.

Table 3.3. Maternal and paternal scores for the four aetiology groups. Means (and SD).

Measure	Maternal outcomes					Paternal outcomes				
	AS	CdLS	CdCS	Autism	Post-hoc	AS	CdLS	CdCS	Autism	Post-hoc
HADS anxiety	11.71 (3.97)	8.93 (4.73)	9.49 (2.90)	9.95 (4.07)	--	10.42 (4.72)	5.85 (4.35)	9.00 (4.16)	7.83 (3.83)	--
HADS depression	8.57 (3.08)	7.30 (5.03)	7.36 (3.42)	6.70 (3.68)	--	8.50 (4.49)	4.29 (2.64)	6.92 (4.09)	5.33 (2.16)	--
QRS-F Family problems	10.61 (2.18)	5.86 (3.38)	7.43 (2.91)	8.37 (3.53)	AS> CdLS**, CdC*	10.24 (2.28)	4.23 (3.52)	7.21 (4.75)	8.00 (2.19)	AS> CdLS**
GSSS	26.31 (8.17)	19.76 (8.78)	20.94 (7.23)	--	--	20.75 (10.07)	16.18 (98.67)	16.68 (5.52)	--	--
Positive Affect Scale	18.64 (6.89)	21.53 (10.84)	19.05 (9.41)	20.60 (7.34)	--	20.33 (9.64)	25.43 (8.37)	20.38 (7.72)	21.83 (5.27)	--
Positive Gain scale	6.93 (3.50)	5.67 (5.49)	7.39 (2.55)	--	--	9.33 (2.77)	5.86 (4.66)	9.38 (4.99)	--	--

* p<.05

** p<.01

Discussion

The present study revealed four general findings: (1) Mothers of children with Angelman, Cornelia de Lange and Cri du Chat syndromes, and fathers of children Angelman and Cri du Chat syndromes were more likely to report clinical levels of anxiety and depression symptoms than normative samples. (2) The likelihood of parents reporting clinical cut off differed between aetiology groups (e.g., both mothers and fathers of children with Angelman syndrome were more likely to reach clinical cut off for anxiety than parents of children with Cornelia de Lange syndrome). (3) Both mothers and fathers of children with Angelman syndrome had the highest levels of negative outcomes, higher even than a comparison group of parents of children with autism. (4) There were no consistent group differences on measures of parental positive well-being. This pattern of results was found even after the groups were selected for the frequency of behaviour problems, which in previous research have been found to be strongly associated with parental psychological distress. In addition, these results were relatively independent of other group differences on child and maternal age, and child adaptive skills.

These results need to be considered alongside a number of methodological limitations. Most notably, the group sizes were very small (especially for fathers) thus reducing statistical power to reveal group differences. However, most results were confirmed using a more stringent analysis, and robust group differences were still evident, despite the low sample size. The mean scores in Table 3.3 indicate that there may well be further meaningful group differences that could emerge given larger samples in future research. In particular, the findings are consistent in indicating that the parents of children with Angelman syndrome reported the highest levels of negative outcomes, even in excess of the scores obtained for parents of children with

autism and an intellectual disability. Small sample sizes are a common difficulty within research on rare syndromes, and efforts to recruit larger samples would be useful in future research.

Additionally, the majority of parents in this study were members of their child's syndrome national support group, and were willing to participate in research. Such participants may represent a particularly well-informed and committed group of parents, and thus the representativeness of the samples is unknown. There was also a lack of confirmatory diagnostic data on children with autism, and a reliance on parental report of their child's diagnosis.

Despite these methodological limitations the findings of the study raise some important questions for future research. In particular, is there something about the behavioural phenotype of children with Angelman syndrome that contributes to highly elevated stress and anxiety levels in parents? Common behavioural features of the syndrome such as short attention span, increased sociability, hyperactivity, aggressive behaviour, and sleep disorder (Clayton-Smith & Laan, 2003; Horsler & Oliver, 2006) may mean that children with Angelman syndrome are uniquely challenging for parents. Behaviours associated with Angelman syndrome such as laughing and smiling are known to increase attention from mothers (Oliver et al., 2007). Although naturally perceived as a positive attribute, it is possible that increased sociability could also cause difficulties for parents. Recent research suggests that the motivation to seek social contact and especially eye contact may underlie aggressive behaviours that function to reinstate adult attention among children with Angelman syndrome (Tunicliffe, 2009). Additionally, raising a child who has a strong and constant desire for social attention is likely to be very demanding for parents. Perhaps focusing on aspects of the behavioural phenotype

such as sociability will help unravel possible aetiology-related causes of the increased parental stress found among parent of children with Angelman syndrome.

Secondly, is there something about the *rarity* of genetic syndromes that contributes to parental psychological distress? In the present study, parents in all three of the rare syndrome groups had mean scores within the mid range of possible total scores on the GSSS. This raises the possibility, that some stressors may be specifically related to the rarity of their child's syndrome. These data suggest that future research into aspects of stress, specifically associated with rarity of syndromes is warranted and qualitative designs might also help to elucidate some of the processes that lead parents to experience these potential stressors.

As far as we are aware, this is the first study to quantitatively measure positive well-being and perceptions of positive gain in parents of children with rare intellectual disability syndromes. The data are encouraging, in that parents of children with rare syndromes all reported positive affect and perceptions of positive impact. There were no statistically significant group differences and the mean scores in Table 3.3 are generally similar, supporting the notion that positive outcomes may be relatively independent of child characteristics (Hastings & Taunt, 2002).

Fathers of children with Cornelia de Lange syndrome reported similar levels of anxiety and depression to the normative population, and the lowest stress levels of all other parents. Given previous reports of high stress levels among mothers of children with Cornelia de Lange syndrome (Richman et al., in press), some elevated negative outcomes for fathers might also be expected (McCarthy et al., 2006). As the group of fathers was small ($n=14$), replication studies are needed to determine whether the current findings are a true reflection of how fathers of children with Cornelia de Lange syndrome adapt to their family situation.

Even given the study's limitations, the findings demonstrate the high degree of stress and the vulnerability to clinical levels of anxiety and depression experienced by parents of children with these rare genetic syndromes, even when compared to parents of children with autism. The results from this study and previous research (Richman et al., in press; Wulffaert et al., 2009) suggest that access to appropriate intervention is essential for parents of these children. In particular, as the prevalence of challenging behaviours is high, early behavioural interventions could be important in minimising the development of challenging behaviours. Ideally, these interventions would take into account the behavioural phenotype of the child's syndrome, and thus be carefully targeted at likely areas of difficulty. Moreover, behavioural intervention at an early stage may lead to a reduction in parental stress, so helping to prevent the mutually reinforcing cycle between child challenging behaviour and parental stress (Hastings, 2002).

Secondly, care providers may be able to anticipate family stress given the behavioural phenotype of the child's syndrome, and thus target parents most likely to require it. Parental interventions should also take into account the genetic syndrome of the child, and emphasise the fact that certain problem behaviours among children with rare syndromes are genetically influenced, thereby reducing parental guilt (Hodapp, 1997). Finally, there is also evidence that the provision of parental workshops targeting parents' own cognitions about their child may help to reduce parental stress (Singh et al., 2006).

Chapter 4. 'You have to sit and explain it all, and explain yourself'. Mothers experiences of support services for their offspring with a rare genetic intellectual disability syndrome

Abstract

Background. Mother's experiences of support services for their adult offspring with a rare genetic syndrome have not been previously explored in research.

Methods. Eight mothers of adults with Angelman, Cornelia de Lange, or Cri du Chat syndrome were interviewed. Interpretative Phenomenological Analysis (IPA) was used to interpret the interviews.

Results. Four themes emerged from the analysis: (i) Uneven medical and social care service provision, (ii) The inertia of social care services, (iii) Mothers as advocates, and (iv) The rarity of their offspring's syndrome.

Conclusions. These findings suggest that accessing appropriate social care services is a lengthy and complex process for mothers. These data may help inform care service providers about how best to support young adults with rare genetic syndromes and their carers.

Research on parents of individuals with intellectual disabilities has largely focused on the childhood years. Quantitative research has suggested that the

increased demands due to a child's personal, medical, and educational needs may place parents at increased risk for stress and depression when compared to parents of typically developing children (Olsson & Hwang, 2001). When parents are asked directly about their experiences of raising a child with an intellectual disability in qualitative research, they identify a number of problematic issues, such as shock and distress around the time of diagnosis of their child (Kearney & Griffin, 2001).

Parents have described having many worries and fears about their development, and, at times, feelings of hopelessness when trying to help their child. As well as this, many parents reported that coping with negative attitudes of others (including professionals) towards their child added an additional burden (Kearney & Griffin, 2001).

Of course, not all experiences are negative and many positive dimensions have also been identified in qualitative studies. Parents were also keen to stress that they felt the experience of raising a child with an intellectual disability had made them better people, and some reported it had made them stronger and more tolerant (Kearney & Griffin, 2001). The considerable positive impact of having a child with intellectual disabilities can occur concurrently with any negative impact (Glidden & Johnson, 1999; Hastings & Taunt, 2002). Experiencing both positive and negative feelings at the same time has been described as a 'tension' for parents, whose joy in their child is also a part of the pain they experience (Kearney & Griffin, 2001).

Generally, less is known about parental experiences in the mid and late stages of their parental career, although many adults with intellectual disabilities continue to live in the family home well into their middle age (McConkey, 2005; Todd, Shearn, Beyer, & Felce, 1993). In the UK, it is estimated that around 50-60% of adults with intellectual disabilities are cared for by their parents (McGrother,

Hauck, Bhaumik, Thorp & Taub, 1999), and around 60 % of people with intellectual disabilities in the United States and 60 % in Australia live with family caregivers (Braddock, Emerson, Felce, & Stancliffe, 2001). Caregiving for an adult is reported to be both rewarding and stressful for parents, and parental experiences of caring for their adult offspring with intellectual disabilities is an area of increasing interest.

When interviewed, parents reported a felt difference between their lives and those of parents of typically developing adult offspring, and felt they were living in ‘frozen animation’ as many caregiving tasks had changed little since their offspring’s childhood (Shearn & Todd, 1997). Their parenting role had been extended indefinitely; some parents found this limiting and reported wanting a life beyond parenting, but the practicalities of taking care of their offspring somewhat prevented this (Todd & Jones, 2005). Additional worries about what would happen to their offspring after their death were also a source of concern (Todd & Shearn, 1996).

There are few studies about the impact of support services on parents of adults with intellectual disabilities. Previous research shows that the use of formal support (as measured by the number of services used out of five) has been found not to be associated with parental quality of life (Walden, Pistrong, & Joyce, 2000). Informal support (provided by family, friends etc) was associated with parental quality of life outcomes. This is inconsistent with the assumption that formal support helps improve parental quality of life (Krauss, 1986).

Although there have been research studies focused on the experiences of parents who care for adults with intellectual disabilities, developments in the field generally relating to genetic syndromes have been rarely considered. There is a growing interest in differences related to genetic aetiology in the wider literature

surrounding intellectual disabilities and the family (Hodapp, 1997). Although parental experiences of caring for children with rare syndromes has started to be explored in small numbers of both quantitative (Abbeduto et al., 2004; Sarimski, 1997) and qualitative studies (Strehle & Middlemiss, 2007), research about the experiences of parents of adults with rare syndromes is scarce and we could find no such studies published in peer reviewed journals. Therefore, the purpose of the current study is to explore maternal experiences of support services for their adult offspring with one of three rare syndromes associated with intellectual disability: Angelman syndrome (AS), Cornelia de Lange syndrome (CdLS), and Cri du Chat syndrome (CdCS).

The three syndromes are each associated with intellectual disabilities and have unique behavioural phenotypes and facial characteristics. All three syndromes are associated with behaviour disorders, such as self-injury and aggression. These significantly impinge on the quality of life of adults with severe intellectual disabilities and their families (Borthwick-Duffy, 1994; Nissen & Haverman, 1997; Konarski, Sutton & Huffman, 1997) and can lead to social exclusion and the need for costly services (Borthwick-Duffy, 1994).

CdLS is estimated to affect 1:40,000 live births (Beck 1976: Beck & Fenger, 1985). The majority of individuals with CdLS have profound (45.6%) or severe (30.43%) intellectual disabilities (Berney, Ireland & Burn, 1999). Health problems are a dominant feature of CdLS, commonly including gastro-intestinal disorders. (Jackson, Kline, Barr & Koch, 1993). Common behavioural features include: anxiety, oversensitivity, sensory self-stimulation, self-injurious behaviour, and compulsivity (Basile, Villa, Selicorni, & Moltini, 2007).

Prevalence for AS is estimated at around 1:10,000 to 1:40,000 live births (Buckley, Dinno, & Webber, 1998; Clayton-Smith, 1993). The severity of intellectual disability ranges from moderate to profound, with severe intellectual disability being the most common (Clarke & Marsten, 2000). Around 80% of individuals with AS have epileptic seizures (Clayton-Smith & Laan, 2003). Frequent smiling and laughing, hyperactivity, and sleep disorder are common behavioural features of the syndrome (Clayton-Smith & Laan, 2003; Horsler & Oliver, 2006).

CdCS is so named because of a characteristic 'cat-like' cry, apparent immediately after birth. The prevalence of CdCS is estimated at 1:50,000 live births (Niebuhr, 1978). The degree of intellectual disability ranges from profound to moderate (Cornish & Bramble, 2002; Sarimski, 2003). Self injurious behaviour and hyperactivity are associated with the syndrome (Cornish, Bramble, & Munir, 1998; Cornish & Bramble, 2002).

Little is known about parents of children with rare syndromes, although there are a few questionnaire-based studies examining parental outcomes (Hodapp, Wijma, & Masino 1997; Sarimski, 1997; van den Borne et al., 1999; Wullffaert et al., 2009) which all find elevated stress and/or depression levels among parents of children with AS, CdLS, and CdC syndromes. Although we know that parents of children are at increased risk of experiencing stress, there is no research on parents of adults with these three rare syndromes, or how their experiences of support services for their offspring may differ from mothers of offspring with more common types of intellectual disabilities. By interviewing mothers who care for adult offspring with CdLS, AS, and CdCS, we sought to describe mothers experiences of support services. Due to the scarcity of research in this area, this study is exploratory in nature.

Method

Methodological approach

As the current research focuses on the previously unexplored population of mothers of adults with rare syndromes, inductive Thematic Content Analysis (TCA: Krippendorff, 1980) has been used, as it is well suited to investigating novel areas of personal experience. Unlike Grounded Theory, which examines social processes and attempts to construct an explanatory framework for the phenomenon under investigation (Willig, 2001) TCA derives concepts from the data. TCA aims to reveal, not inhibit, the diversity and richness of individuals' experiences within similar situations and the interviewee is regarded as an expert in their own experiences. It is a 'bottom-up' approach, and the data are coded without trying to fit them into a pre-existing theme (Elo & Kyngas, 2007).

A key process in TCA is the dynamic interpretation of the interviews, as the researcher is active in conducting the research and in interpreting the participant's responses. It is recognised that researcher interpretative activity is inherent in the method of TCA, and that the researchers' own preconceptions will necessarily influence this process (Graneheim & Lundman, 2004; Krippendorff, 1980). In the current study, the primary researcher was a research student with interests in parental adjustment to having a child or adolescent with CdCS, AS, or CdLS, and who had previous experience of interviewing parents of children with intellectual disabilities over the telephone for a quantitative study focused on these same syndromes.

Participants

Eight mothers of adults with rare syndromes were interviewed. The ages of mothers ranged from 51 years to 72 years ($M= 55.87$ years, $SD= 6.75$), and their offspring ranged from 24 years to 44 years of age ($M= 29.33$ years, $SD= 6.18$).

All participants were biological mothers, and all were married and living with their husbands, and had no other children with an intellectual disability (apart from one mother who had two daughters with AS). Three mothers had sons with a rare syndrome, and five had daughters. Two mothers had offspring with CdLS, three had offspring with AS (one of these mothers had two daughters with AS), and three had offspring with CdC syndrome. All names and places in the present paper have been changed or deleted to protect the identity of the participants. See Table 4.1 for details on individual mothers and their offspring.

Procedure

The mothers originally responded to a mail shot campaign recruiting participants for a wider research study about challenging behaviour in children and adolescents with CdLS, AS, and CdCS. However, because their offspring did not meet the criteria for the study (because they were over the age of 19), they were invited to take part in the current study.

Table 4.1. Characteristics of mothers and their offspring, current living status and services received.

Mother (age)	Young adult (age)	Diagnosis	Current living status and services received
Natalie (55)	Sarah (25)	Cri du Chat syndrome	Lives in family home; goes to day centre on weekdays. Does not use respite services.
Grace (72)	Chris (44)	Cri du Chat syndrome	Lives in family home; goes to day centre weekdays and uses respite services.
Olivia (52)	John (24)	Cri du Chat syndrome	In supported living full time.
Megan (52)	Ross (27)	Cornelia de Lange syndrome	Lives in family home; goes to day centre weekdays and uses respite services.
Joanna (55)	Julia (30)	Cornelia de Lange syndrome	Lives in family home; goes to day centre weekdays and uses respite services.
Katie (56)	Holly (32)	Angelman syndrome	In supported living full time.
Helen (54)	Lisa (27)	Angelman syndrome	In supported living full time.
Sophie (51)	Charlotte (30)	Angelman syndrome	In supported living full time.
	Ellie (24)	Angelman syndrome	Lives in family home; goes to day centre weekdays and uses respite services.

An information pack containing a cover letter, information leaflet, and consent form were posted to the mothers (see Appendix 18). Of the ten mothers contacted in this manner, seven consented to take part in the study. One mother was also recruited from a syndrome support group meeting, in which the same information packs about the study were handed out. The only inclusion criterion was that they were the primary carer of an adult (aged over 19) with, CdLS, AS, or CdC, and there was no upper-age limit for participating in this research.

Once the mothers had returned the consent form with their written consent, a researcher contacted them to arrange a suitable interview time and to answer any questions. Interviews were conducted over the telephone rather than face-to-face to facilitate participation, as the participants lived in various geographical locations across the UK. Interviewing via telephone has been affirmed as a useful method for conducting qualitative research (Sturges & Hanarahan, 2004), and has been used successfully in IPA research as a method of interviewing parents of individuals with intellectual disability (Reilly, Huws, Hastings, & Vaughan, 2008).

In addition to written consent, verbal consent to record the interview was gained just before the interview commenced, and all interviews were recorded on a digital recorder. They lasted from 54 to 96 minutes (Mean = 76.12 minutes) and were one-time interviews. A semi-structured interview was developed for the purpose of this study and it included subsidiary questions and prompts. The interview started with general demographic questions to help the mothers become familiar with the researcher and to feel more at ease. The interview was designed to explore parental experiences of social services for their adult offspring with a rare syndrome. The interview involved questions about current social care services received and their opinions of it, medical services received, encounters with

professionals, and thoughts about future care for their offspring. The interview also explored whether mothers had encountered any problems with services attributable to the rareness of their offspring's syndrome (see Appendix 19). These questions were flexible to allow pursuit of any topics that arose during the interviews which had not been identified by the researcher. Mothers did not see a copy of the semi-structured interview, but did receive a general outline of the topics to be explored in the information sheet provided.

Data analysis

Firstly the recorded interviews were fully transcribed by the researcher. The process of transcription helps the researcher become familiar with the interviews, and some regard it as a key phase of data analysis (Bird, 2005). The researcher then conducted open coding, where the transcriptions are read through line by line, noting points of interest or significance on the transcript in the left hand margin. The readings were repeated and emerging themes were noted on the right hand margin. This was repeated until the researcher was satisfied that all possible categories, or 'themes' had been identified. These themes and supporting quotes from the interviews were compiled in higher order headings in a separate document, and connections between them were noted. This process was repeated for all eight transcripts. The themes from all the interviews were then compiled and compared, producing a list of 'master' themes which best represent the interviews, along with emerging sub-themes (Elo & Kyngas, 2007; Graneheim & Lundman, 2004; Krippendorff, 1980). To ensure validity of these themes, this process involved other researchers reading each transcript and developing emergent themes until all were satisfied that data saturation had been achieved. Master themes were then developed

via discussion, which ensured that the themes were grounded in material from the transcripts. Additionally, to ensure the validity of the themes, the results section was posted to all participants with an invitation to comment if they so wished (see Appendix 23). Two mothers responded with requests for the paper if published but did not make comments on the themes.

Results

The four master themes which emerged were: (a) Uneven medical and social care service provision, (b) The inertia of the social care system, (c) Mothers as advocates, and (d) The rarity of their offspring's syndrome.

When providing extracts from the interviews the following conventions are used:

... Short pause

[text] Explanatory information provided by author

(...) Words omitted to shorten quote

Theme 1. Uneven medical and social care service provision

Mothers reported widely different experiences of medical and social care provision for their offspring, although they were largely negative. Within medical services for example, half the mothers reported that they had experienced a form of prejudice against their offspring. One participant believed that the reason her son's health care was inadequate was due to a '*Medical system that doesn't allow for disability*' (Olivia), whereas Natalie believed that within the medical system '*Our kids are treated as second-class citizens*'. Olivia felt the diagnostic process for her son was impeded by faults in the medical system. Over a period of five years, her

son, John's weight dropped to around 38 kilograms, and Olivia went to an enormous effort to get the medical consultant to investigate her son's health thoroughly.

Olivia: So with the letter from the social worker going to the consultant, with the letter from the college going to the consultant, with my own doctor's letter going to the consultant, and with the learning disability nurse coming along with us to the appointment - they started to look a bit closer.

After this effort, the consultant diagnosed John with hyperthyroidism. The length of time it took to get this diagnosis was felt to be a subtle form of prejudice that was inherent in the medical system, as the consultant did not take John's health seriously. A further barrier to accessing healthcare was some staff's lack of experience with people with intellectual disabilities, and the quality of care received was largely dependant on the staff members who worked directly with their offspring. At times, mothers found themselves teaching healthcare staff about intellectual disabilities, which was both frustrating and time consuming.

Sophie: Well if they [medical staff] are involved in disabled people they are good (...) But if you go to other places that don't deal with disabled people it can be different, 'cos then you have to sit and explain it all and explain yourself.

Additional medical complications surrounding intellectual disability are not always addressed sufficiently. Sophie's daughter, Charlotte was diagnosed with polycystic ovaries, but if given any medication containing artificial colours her challenging behaviour is likely to increase for around six months. The medical consultant's advice on this matter seems wholly inadequate:

Sophie: Well they just said to me 'Go to your chemist and ask them what is in it [the medication] and find out - and then if it's too much trouble for yourself to handle it, because of her behaviour, then leave it out'. That's the answer.

Some mothers felt that medical staff's lack of experience and expertise contributed to a lengthy diagnostic and/or treatment process for their offspring, and that this was unacceptable practice.

Widely different experiences of social care provision were reported by mothers. Appropriate day centre or living placements were scarce or non-existent, so most of the mothers reported having to take what was available in their local area and '*making the best of it*' (Katie). The most pervasive problem with day, respite, and residential care services was the frequent turnover of care staff who worked directly with their offspring, which created multiple problems. Mothers believed that high staff turnover led to challenging behaviours and a reluctance on the part of their offspring to attend day and respite placements.

Joanna: The trouble is though the turnover of staff is so...regular (...) she gets to know them and then they leave. Then it's like starting over again, from scratch because she finds it very, very difficult. Anything new, it takes her a long time to get used to.

Staff working in residential care services were often regarded as unreliable and mothers did not trust them to deliver the agreed service.

Katie: They listen to what I say and put it down on paper, and we have action plans and then it doesn't materialise and then we have another hiccup and the action plans fall by the wayside.

These situations occurred frequently and led to many mothers being uncertain about the quality of support their offspring received from residential services and

day centres. Because they did not trust the day or residential care providers, many mothers felt they had to get deeply involved with them to ensure their offspring's well-being. Sophie's daughter Charlotte has been in the same supported living placement for six years, during which time it was run by three different companies. The staff turnover was so high that often they were not trained, and sometimes were not even competent.

Sophie: They [staff employed by the first care provider organisation] didn't know how to cook, they didn't know how to wash her clothes. They dress her in summer clothes on a winter's day or the reverse way round, there was so many problems, they stole her money the first week she was there. They kept having to change the staff, I gave complaint after complaint after complaint until they got rid of them. And then they left and they got another company in and they were there for two years and I fought tooth and nail with them because of the care.

In addition to dealing with constant problems with day or residential care services, two mothers reported that their offspring had experienced physical abuse from social care staff. Megan's son, Ross came home from the day centre with bruises and he and other service users were the subject of a thorough police investigation, with two staff members being removed from their jobs as a result. Natalie's daughter, Sarah, had been physically abused as a teenager on two occasions by care staff working at respite services. Sarah suddenly became reluctant to go to respite, and after some careful questioning by Natalie, indicated that a member of staff had been physically pushing her hand up behind her back.

Natalie: She actually showed me what they did to her. So we took it further but because Sarah, with her speech, we were basically, we were told it was

her word against this other persons, so basically you haven't got a leg to stand on have you?

Although this should have been the basis of an investigation by police and social care services, Natalie was unable to take it any further due to her daughters difficulties with communication. This experience left both Megan and Natalie with deep reservations about the quality of staff and the standard of care provided by social care services.

In contrast, three mothers reported receiving good day or residential services at times, although only one mother was totally satisfied with her offspring's residential placement. Helen's daughter, Lisa had been living in the same supported living home for eight years; the grounds of the home were attractive and well kept, and there was a low turnover of staff. If there were ever any issues (for example when Lisa got hit by another service user) both Lisa's parents and social services were immediately informed, which Helen found reassuring.

Helen: She always looks well, she looks happy, she wants to go back. You know she loves coming home, and she just slots in as though she has never been away, but you take her back and they say the same.

Mothers' most commonly reported area of satisfaction was that day centres enabled their offspring to experience a varied and full life.

Natalie: I wouldn't mind going [to the day centre] myself. And she has a brilliant time she does horse riding, swimming, bowling, yachting, canoeing, she does all sorts, every day she's out. It's just up Sarah's street, she loves it.

Likewise, three mothers felt they had received good medical care for their offspring over the years.

Grace: We've never had any problems when we have turned up with him anywhere, and I know some people do, but locally here, we seem to be ok really.

There appears to be an uneven quality in health and social care service provision received by these mothers, with the majority of mothers reporting a poor standard of care, and felt this was often due to the high turnover of support worker staff who worked directly with their offspring. However, it is important to note that a minority of mothers did feel their offspring were receiving a good standard of care from social care services. This was often seen as an atypical situation, and parents frequently described themselves as 'lucky' to receive such provision. Likewise, Todd and Jones (2003) found that parents felt that to receive a high standard of social care was an unusual event, and also attributed this to luck.

Theme 2. The inertia of the social care system.

The administrative system of social care services was regarded as complex and mothers had to work hard to ensure their offspring's support needs were met. Staff working for social care services seldom responded to requests unless repeatedly chased by mothers. Mothers rarely talked about individuals within social care services as being difficult or causing problems, it was the organisation of a faceless 'system' that they perceived as being problematic and inert. Social care services were often referred to in the third person (e.g. 'them' 'they') and this reflected a felt separateness. There was also little parental report of a collaborative effort between themselves and social care services to support their offspring. Thus, in addition to negative or positive day-to-day experiences with those who worked

directly with their offspring, the administration side of the social care system was perceived as rigid.

Natalie: Social services is a waste of time. The excuse you get as well that they are short-staffed, it's this it's that it's the other. They've got a lot on, and you just basically think to yourself "I think I'm wasting my time here" and I think that's what they hope you're going to do, that you're going to give up in the end, and you do, inevitably. You get cheesed off with waiting for them to call you and you calling them.

Although a role of social workers is to help parents access the services they need, getting and retaining a supportive social worker was difficult. The majority of mothers had had many different social workers over the years, and two mothers at the time of interview did not have one at all, and were told there were none available.

Megan; You just get used to one, you fill out all the paper work "That's fine" they say "We'll go away we'll do this, that, and the other" ...Silence. So then we try and find out what's going on and they say "Oh sorry that social worker's left and there's nobody in post" So you get those sorts of problems.

Even at the top of the staff hierarchy at social care organisations mothers experienced problems. Joanna found that the managers of the local intellectual disability team had little experience with people with intellectual disabilities, and as a result had many misconceptions.

Joanna: A few of the top people of the social services and [name of] County Council have never come across or never dealt with disabled people. And they are put in these positions without realising what they are going to do and they expect half of them [people with intellectual disabilities] to be able to work,

and they can't work. (...) They don't want to know, and I'm thinking 'You've got all these top jobs.' What are they doing?'

Problems with the hierarchy within social services were also reported by Olivia. She found that during meetings with social care services, no single person takes control of making a decision and ensures it is followed through; this slows down the decision-making process. A lack of an expediter is seen as a fundamental difficulty when communicating with social services.

Olivia: That is the whole problem with statutory services, they have no word for expedite. They just don't understand about moving things on, and that is the whole problem with everything, (...) whatever I've come to, nobody has had the responsibility of making it happen, they all make judgements, but nobody is actually making sure it happens.

As well as mothers reporting difficulties with the organisational structure of social care services, trying to get basic day-to-day questions answered by social care services was problematic. Despite some mothers having received services from their local authority for over a decade, they still receive an indifferent response when they try to communicate with administrative staff working for local social services.

Joanna: Whenever I ring up I always speak to somebody different, you never speak to the same person ... but you go and ring somebody up now and ask them a question they will say "Does she have to have help? Does she need help?" Well would I be ringing if I didn't? I mean I wouldn't bother to ring would I?

All mothers interviewed found communicating with social services problematic, and were aware of other issues besides the well-being of their offspring

which influence care workers' decisions. Distrust of social care services was implicit throughout many of the interviews

Olivia: The issue is they don't have one agenda do they? (...) They might have my sons' interest at heart, but they also have the budget at heart and keeping their jobs at heart, and a lot of other things are influencing their decision as to how good my sons' care is.

Theme 3. Mothers as advocates

Mothers often referred to attempts to access appropriate social care for their offspring as 'fights' and a sense of 'us' versus 'them' emerged from the interviews. Mothers were forced into a role of advocate for their offspring, because if they did not advocate, they felt their offspring would receive sub-standard social care.

Katie: The services don't come to you, and you don't get the changes unless you are out there fighting, which isn't the right thing to be doing really, you don't want to be fighting for everything you get, but sometimes it feels like that.

Throughout the interviews mothers described having to 'fight' or 'battle' with social care or medical services to receive anything beyond minimum provision. Placements that were first offered by social services were largely found to be unsuitable – for example it was suggested that a residential home for the elderly would provide respite care for Joanna's 30-year old daughter. This was totally unacceptable to Joanna, so she then had to 'fight' this in order to get suitable respite care. Social care services would only respond to mothers' requests if they repeatedly and assertively 'fought' for an appropriate care service.

Katie: You get your basics but you know if you're wanting anything that you think is what should be acceptable for her [my daughter], then you have to have a battle about it, you have to have a battle.

Mothers needed to be persistent in their communications with social services, and could not rely on anyone else (including social workers) to get acceptable services for their offspring. Thus, mothers were forced into a role of being advocates and 'fighters'. This seemed to be a large part of their identities as mothers, as the role of advocate was at a level of involvement not usually experienced by parents of typically developing offspring.

Helen: You have to fight all the way. You have enough of a fight often just with day to day living. And if you let things get on top of you it can be really difficult.

Mothers often felt as though they were not listened to and so were forced to go to a higher authority to get suitable services. In all, seven out of the eight mothers had reported doing this. Four mothers had contacted their local Member of Parliament (MP) about getting services for their offspring. This was done after mothers had tried and failed to get access to appropriate services for their offspring via standard routes.

Olivia: The only time things started to change was when I said to the MP, and said look, can you help me? And once ... the awful thing is that it takes that, for people to be doing anything.

Another parent threatened legal action against their local social services, another went to a local legal tribunal, and another went to the High Court with around 50 other families to protest against her local council trying to close all respite and day services within her local area. Some mothers felt conflicted about their role

as an advocate, and felt that they were a nuisance when asking for acceptable services for their offspring. However, they also felt they had little choice but to fight on behalf of their offspring.

Megan: You do feel guilty at sometimes having to shout a bit loud, whereas normally I would sit back and let it go, but you have to be the spokesperson, don't you?

Interviewer: So what would happen if you did sit back and not say anything?

Megan: Probably it would go all to pot really.

The frustration and negative psychological impact of dealing with the network of social services was apparent throughout most of the interviews.

Natalie: We never had any help, it was always a fight all the time to get anything done. Over the years you get so tired of getting your knickers in a twist all the time. When you ask nicely, nothing gets done. It was the Mums at the school used to say "Natalie you have to get nasty because that makes them listen". And that proved to be correct. But it does get tiresome.

Some mothers reported that the stress of advocating for their offspring affected their health, some attributing high blood pressure or nervous breakdowns to the additional burden of constantly making sure their offspring was cared for appropriately by social care services. At the time of the interview, Sophie had been waiting for eight months for a suitable supported living placement for Ellie, her younger daughter.

Sophie: I can't manage myself I know, I can't carry on. I want to give it up [the care] because of my three breakdowns and I want my life back. I've had 30 years of it and it was easier when they were children...you think it isn't, but it was.

Clearly, involvement with social services added a major dimension into these mothers' lives throughout their offspring's adult years, a dimension which was largely negative and frustrating. Some mothers found that their role as 'fighters' was made easier by becoming involved with the social care system in other ways, such as being a parent representative on local intellectual disability advisory boards or working for intellectual disability charities. These proactive strategies involved a lot of commitment but mothers felt they were more likely to be listened to by social care services as a result.

Grace: I've been for years a campaigner of some form or another, (...) when you say what your name is they do know, you know, who it is. And I think you have got to be very vocal and just keep on, just keep on making yourself heard all the time.

Some mothers felt that a reputation of determination and persistence helped them be listened to by social care services; but it took years of hard work to get to this point.

Olivia: Am I listened to by services? Yes I am now, I am now, but that is only because half my life is spent...and they know that we are helping them. It's not because they love me. But I would say the only reasons I am listened to is because they know that if they are not listening to me, somebody else will be.

Theme 4: Rareness of syndrome

Very few participants' spoke spontaneously about the rareness of their young adult's syndrome and the impact of this on the services they received. This may reflect the fairly neutral attitude expressed about the rarity of their child's syndrome

in relation to services, as 7/8 mothers felt the rareness of their young adult's condition did not directly affect their access to services.

Grace: They're just all lumped together as a learning disability and nobody really knows (...) I mean it doesn't bother us in the least if he is lumped in with other people with learning disabilities, because he has a learning disability and therefore you know we just live with that.

Mothers found that their offspring were categorised as having an intellectual disability by social services, with little recognition of their rare syndrome or issues associated with it. However, this was not regarded as a problem by mothers, who felt they had the same opportunities to access services as mothers of adults of any other type of intellectual disability. A desire or need for any specialised services for people with rare syndromes was not expressed during the interviews. It seemed that the mother's primarily identified themselves as being mothers of a person with intellectual disabilities, and the rare genetic syndrome of their offspring became a secondary issue. This was certainly the case for Katie, whose daughter Holly had a late diagnosis of Angelman syndrome at the age of 23. She felt the diagnosis had little impact on her perceptions of her daughters' disability, but speculated on whether an earlier diagnosis would have made a difference to the way she approached teaching Holly communication skills.

Katie: I can't say it has made any difference because she's got Angelman's syndrome, she's just a child that has got severe learning difficulties... I don't think anything would have changed within that, apart from I felt the communication, which may have been handled differently.

Some mothers felt that obtaining health care for their offspring was made more difficult because of the rareness of their offspring's syndrome, as some

medical professionals have little knowledge about rarer syndromes and as a result may not be able to give specific medical advice.

Natalie: They won't admit to you they don't know much about Cri du Chat.

(...) And they are asking me questions about Cri du Chat, and I'm saying

"Are you not supposed to be telling me? You're the professionals, I'm just a Mum".

Additionally, the rarity of a syndrome means that there is often very little research and information about how the syndrome may affect people during their adult life, and the average lifespan is unknown. This lack of knowledge left some participant's feeling unprepared and unsure about the future.

Sophie: If you knew their lifespan you would know what to expect and which road you're going to go down. It's not fun living in the dark and that's what I've lived in for 30 years, not knowing what is the next stage.

Natalie: Basically it's the blind leading the blind if you like, not knowing what to expect what the future holds.

Caroline and Sophie's similar use of metaphor in the above passages ('blindness' and 'living in the dark') serves to emphasise their sense of isolation and uncertainty, and consequential difficulty in planning for the future.

Discussion

These mother's accounts of their experiences of support services highlight a number of important dimensions. Mothers reported largely negative experiences of social care services, similar to those reported in previous research (Shearn & Todd, 1997). They described the difficulties encountered when trying to get acceptable social care for their offspring in day, respite, and residential services, having to

become an advocate for their offspring, and the frustration and stress experienced in this role. Additionally, the influence of the rareness of their offspring's syndrome within social care services was discussed.

Uneven social care and medical services

Although social and medical services are intended to bring positive benefits for service users and their families, the current research suggests that any benefits are often coupled with effort and frustration when trying to access support services. Mothers want good services for their offspring, and not simply the bare minimum. It is here the conflict arises between mothers and social care providers. Mothers described their role as advocates as one of having to 'fight' social care services rather than having a collaborative partnership, and this is consistent with earlier research (Shearn & Todd, 1997; Todd & Jones, 2003).

On a more positive note, two mothers (Grace and Helen) reported that overall, they were satisfied with the support they had received from both medical and social care services. Like other mothers, both acknowledged that negotiating the social service system was difficult and required much involvement, and there was no noticeable difference between their dedication and knowledge of social services to that of the other mothers interviewed. So why did these mothers report satisfaction with social care services whereas others did not? One possible explanation, which was alluded to by several mothers, is that the quality of local social care services is dependent on the standards of the local authority, and is therefore inconsistent throughout different areas in the UK. This difference in social care provision is evident even among the eight mothers interviewed.

Interestingly, whether their offspring lived away from home or in the family home did not seem to affect mothers involvement with care providers, what was influential was the perceived quality and reliability of support services. The less confident mothers were in the quality of care, the more contact they had with the support provider, thus the quality of care influenced mothers responses, not type of care.

Overall, support service provision did not seem to be based on a sensitive understanding of parent's lives. Services need to acknowledge the extra burden they can place upon parents, and a greater priority should be given to monitoring how parents feel about support services for their offspring to re-address the balance towards considering the family of the service user. This could be achieved by ensuring that parents themselves feel supported, and establishing opportunities for parents to express their experiences and expectations of support services.

Parental experiences of medical services for their adult offspring with intellectual disability have not before been directly explored in qualitative research, although health discrepancies between people with and without intellectual disabilities are well documented (Krahn, Hammond, & Turner, 2006). Health service provision for people with intellectual disabilities is known to be inconsistent (Lennox & Kerr, 1997). This was reflected in the current research, with some mothers reporting satisfaction with the medical care their offspring had received, whereas the majority of mothers reported a felt prejudice towards their offspring and reported difficulties in accessing appropriate medical care. The inexperience of some medical care staff and a failure to recognise complications arising from intellectual disability were issues highlighted by mothers. A need for specialist training and

guidelines for medical staff is needed to assist them in best to addressing these issues.

Whether the problems that mother's experience in relation to support services change as their offspring gets older was not specifically addressed in this study. However, there were no noticeable differences in the involvement of the mothers, or how they perceived services although there was a substantial age difference in their offspring (age range 24-44 years). Again, it was the quality of services which influenced mothers' responses above all other factors.

The rarity of their offspring's syndrome

The experiences of support services which emerged from the analysis are unlikely to be unique to mothers of adults with rare syndromes, as their accounts have much in common with mothers of adults with other types of intellectual disabilities (Shearn & Todd, 1997), and seven out of eight mothers felt the rareness of their offspring's syndrome did not affect their access to social care services.

However, the lack of knowledge about a syndrome has a two fold impact on mothers. Firstly, mothers felt 'in the dark' were unsure as to what to expect during their offspring's adult life. Secondly, it may impede access to appropriate medical care as some professionals may not know about the syndrome.

Mothers felt that they had the same opportunities to access social care services as other parents, and the rareness of their offspring's intellectual disability made little difference in their relationship with social care services. However this may reflect the lack of awareness of the importance of syndrome-specific issues by many professionals, rather than the diagnostic status being of little importance in itself. It is clear that more research into older individuals with rare syndromes is

important so as to increase the availability of information to parents, as not knowing developmental outcomes, such as the potential life-span of their offspring, caused some mothers considerable worry.

Methodological limitations

There are some methodological limitations of the current study which need to be considered. The small sample size of this study makes it difficult to construct generalizations from the findings and how representative these are of mothers experiences is unknown. Additionally, all mothers are members of the relevant rare syndrome support group, and had responded to a research recruitment leaflet. It is possible that their experiences differ to those parents who are not involved in support groups or who are unlikely to respond to research. Thus, the sample may be biased in the sense that the mothers are willing to participate in research and thus may represent a particularly well-adjusted group. However, given that the mothers reported similar experiences to mothers in previous research (Todd & Shearn, 1995) this study is likely to be a good reflection on how mothers of adults with rare syndromes experience support services.

The interpretative nature of the research was influenced by the researcher's perspectives. This was counter-balanced with efforts to ensure that the interpretation was fully grounded in the data, by discussions about the analysis and subsequent themes with the second and third authors.

Additionally, some of the mothers recounted past as well as current experiences when describing support services. For the most part, the interview focused on current experiences of social services, but some mothers did recount some past experiences when asked specific questions on the positive and negative

aspects of support services and professionals. So although not all of the interviews were solely about the present experiences of mothers, it could be argued that the overall opinions of mothers are based on their past and present experience, and thus past experiences are relevant as they influenced mothers opinions of support services.

Although relatively small in scale, this study supports earlier research on the difficulties mothers experience in relation to services (Shearn & Todd, 1997). There is an ongoing need to take into account how parents regard the quality of services to help policy makers bridge the gap between the standards of parents and those of services. Furthermore, acknowledging the day-to-day struggles that some mothers encounter with social care and medical services is important for policy makers, as additional stress may compromise some parents' ability to continue caring for their offspring at home. An appreciation of the additional burden that social care services can bring to parents is needed in order to facilitate a mutually beneficial collaboration between parents and service providers.

Chapter 5: Predictors of positive and negative measures among mothers of children with rare genetic syndromes.

Abstract

Background. Few research studies have considered adjustment among parents of children with rare syndromes, although there is evidence that some parents report very elevated levels of stress (Wullfaert et al., 2009). Specifically, little is known about variables that affect parental stress in this population.

Method. Mothers of children with various rare syndromes (n=145) completed postal questionnaires about their child's characteristics (challenging behaviour, mood, health, adaptive behaviour) and negative and positive measures of psychological well-being, including a measure on stressors related to the rareness of their child's syndrome.

Results. Positive child mood was the most frequent predictor of maternal negative and positive measures, whereas child challenging behaviour was only predictive of general parenting stress. Additionally, child age negatively predicted stressors related to the rareness of their child's syndrome,

Conclusions. The findings suggest that child characteristics contribute to parental stress and well-being. This research may help practitioners to identify parents potentially at risk of high levels of stress given the characteristics of their child.

Despite a healthy research literature on both the challenges and positive aspects of raising a child with intellectual disabilities on the family (e.g., Blacher & Baker, 2006; Hassell & Rose, 2005; Hastings & Taunt, 2002; Hatton & Emerson, 2003), the experiences of families of children with rare genetic syndromes have not been widely researched. At the time of writing (December 2009) a systematic search identified fewer than 30 published studies focused on parents and families of children with rare genetic intellectual disability syndromes (see Chapter 1). These few research studies consistently show that parents of children with rare genetic syndromes are more likely to report elevated levels of stress than parents of children with other, comparatively well researched aetiologies. Both mothers and fathers of children with rare genetic syndromes report higher stress levels than parents of typically developing children (Perry, Sarlo-McGarvey, & Factor, 1992; Sarimski, 1997; 1998; Von Gontard et al., 2002), parents of children with a mixed aetiology intellectual disabilities (Hodapp, Dykens, & Masino, 1997; Von Gontard et al., 2002), and parents of children with Down syndrome (Abbeduto et al., 2004; Fidler, Hodapp, & Dykens, 2000; Lewis et al., 2006; Richman, Belmont, Kim, Slavin, & Hayner; in press). There is also evidence that parents of children with rare genetic syndromes report slightly less stress and similar levels of depression to parents of children with autism (e.g., Abbeduto et al., 2004; Lewis et al., 2006), who are themselves widely found to report the highest levels of stress when compared to parents of children with other disabilities (Eisenhower, Baker, & Blacher, 2005; Kasari & Sigman, 1997; Olsson & Hwang, 2001; Sanders & Morgan, 1997).

Although researchers have compared parents of children with rare syndromes to parents of children with other aetiologies, other research questions have received less attention. In particular, there are few current data on the variables that predict

psychological well-being in parents of children with rare syndromes. In existing research, two child behaviour domains have been explored: levels of behaviour problems (e.g., Briegal, Schneider, & Schwab, 2006; Hodapp, Wijima, & Masino, 1997; Lewis et al., 2006; McCarthy, Cuskelly, van Kraayenoord, & Cohen, 2006; Richman et al., in press; Wulffaert et al., 2009) and to a lesser extent, child adaptive behaviour (e.g., Hodapp et al., 1997; Laurvick, Clerk, & Bower, 2006; Perry et al., 1992; Sarimski, 1997). In general, child behaviour problems are positively associated with negative parent outcomes, although some studies have not found this association (Lewis et al., 2006; McCarthy et al., 2006). Similarly, lower levels of adaptive behaviour are associated with higher parental stress, albeit to a lesser extent than child behaviour problems (e.g., Hodapp et al., 1997; Sarimski, 1997).

Variables other than child problem and/or adaptive behaviours have rarely been explored both in the wider research literature related to children with intellectual disabilities and in research specific to families with children with rare genetic syndromes. For example, we could find only two previous studies in the wider research literature where child pro-social behaviour was explored as a predictor of parental well being. These studies found that pro-social behaviour was negatively predictive of maternal stress cross-sectionally (Beck, Hastings, Daley, & Stevenson, 2004) and longitudinally (Neece & Baker, 2008). Similarly, the child's current mental health has been neglected in intellectual disability family research. Until recently it was difficult to assess mental health in individuals with severe or profound intellectual disabilities due to the reliance on self-report to determine a person's mood and mental health. However, there has been a recent interest in depressive symptoms among this population, and some measures have been developed to begin to explore this (Ross & Oliver, 2003a). Although there is evidence that the mental health of

mothers and fathers are predictive of stress levels in their partner (Hastings et al., 2005), this research has not been extended to determine whether the mental health of a child with severe or profound intellectual disabilities may be associated with parental well-being.

In the current research, our aim was to expand the literature on rare genetic syndromes by investigating a broader range of child variables, and to explore both positive and negative maternal outcomes. Thus, in addition to adaptive and problem behaviours, we included measures of the child's general mood, positive behaviours, and, given the increased risk of health problems among individuals with intellectual disability, the child's physical health status (Jansen, Krol, Groothoff, & Post, 2004). In addition to parental psychological distress (stress, mental health problems), we extended this measurement to include stressors that may be unique to having a child with a rare genetic syndrome, and also to positive impact.

We could only find one existing study which explored positive impact upon parents and the family when a child has a rare genetic syndrome. Poehlmann, Clements, Abbeduto, and Farsad's (2005) qualitative study found that all mothers of children with fragile X syndrome (n=11) and all but one of the mothers of children with Down syndrome (n=10) were similarly positive when describing their children. These mothers emphasized qualities such as their child's sense of humour, cheerfulness, caring nature, and ability to connect with family members and friends. Although the generalisability of these findings to other parents of children with rare syndromes is unknown, these data suggest that mothers of children with rare syndromes and Down syndrome may report similarly positive perceptions of their children.

Stressors that parents may experience as a result of the rareness of their child's condition (such as medical professionals having little knowledge of a syndrome, or having to explain their child's syndrome to new people) have not been addressed in any published family adjustment literature. The research in Chapter 3 was the first to explore stressors related to the rareness of the syndrome, and found that these types of stressors were salient to parents, as parent's mean scores on the GSSS were in the mid range of total possible scores. There may be unique stressors attributable to parenting a child with a rare syndrome of which researchers and practitioners are unaware

Given previous research findings, there were two general predictions for the current study: (1) Child problem or challenging behaviour would emerge as a strong predictor of maternal distress, consistent with previous research among families with children with rare syndromes (Briegel et al., 2006; Sarimski, 1997), and (2) Few, if any, child variables would emerge as predictors for positive measures of parental well-being. Due to lack of previous research on child variables such as physical health, mental health, and pro-social behaviour, there were no specific predictions about these child variables. Similarly, again due to lack of previous research on the subject, no hypotheses were made about likely predictive variables for stressors associated with the rareness of a child's syndrome.

Method

Participants

The total sample consisted of 145 mothers (137 biological mothers, five foster mothers, two adoptive mothers, and one grandmother who was the primary caregiver of the child) of children between three and 16 years of age with a diagnosis of one of seven rare syndromes: Angelman, Cornelia de Lange, fragile X, Prader-Willi, Lowe,

Smith-Magenis, or Cri du Chat syndrome. Diagnosis was based on parental report as we did not have access to clinical records. Child age, gender, maternal age, income and marital status for the sample are summarised according to syndrome group in Table 5.1.

Measures

Ten measures were included in the present research. Five looking at maternal stress and well-being, and five examining child variables. A short demographic questionnaire was also included to determine the child's age, gender, and diagnosis as reported by the mother, and the mother's age, marital status, and annual income (see Appendix 20).

Maternal measures

General maternal stress related to having a child with a disability was measured using The Parent and Family problems subscale from the Questionnaire on Resources and Stress – short form (QRS-F: Friedrich, Greenburg, & Crnic, 1983). Five items were removed from the original subscale as they have been identified as measuring depression rather than parental stress (Glidden & Floyd, 1997). Due to limited space in the questionnaire, the remaining 15 items were then reduced to 7 items by selecting the items with the highest item-total correlations using the data from the QRS-F from the participants in Chapter 3. The subsequent scale (see Appendix 21) included the items “1-Caring for N puts a strain on me,” “2-Other members of the family have to do without things because of N,” “3-In the future, our family's social life will suffer because of the increased responsibilities and financial stress,” “4-I can go to visit friends whenever I want,” “5-There are many places where

we can enjoy ourselves as a family when N comes along,” “6-Members of our family get to do the same kinds of things other families do,” and “7-The constant demands to care for N limit my growth and development.” Using this seven item scale (QRS-F7), parents were asked to circle either “True” or “False” for each item based on whether the item applied to their family. There was a strong correlation between the full 15-item scale and the shortened 7 item scale, $r=.75$, $p<.05$. The Kuder-Richardson coefficient (equivalent to Cronbach’s alpha for scales with dichotomous items) for the QRS-F7 in the present sample of mothers was .82.

The Genetic Syndrome Stressors Scale (GSSS, see Appendix 13) was developed for this thesis (see Chapter 3 for details). It is designed to assess parental stressors relating to rare genetic disorders. Parents are asked to rate statements such as “People staring when I go out in public with my child” and “Going to see professionals who are not knowledgeable about my child’s genetic syndrome” on a 4-point scale ranging from “0-not at all stressful” to “3- Extremely stressful.” The GSSS has moderate correlations with both negative and positive measures, and has good face validity and internal consistency (see Chapter 3). Cronbach’s alpha for the present sample of mothers of children with rare genetic syndromes was .87.

Table 5.1. Demographic information about the children and mothers.

Demographics	Angelman syndrome (n=33)	Cornelia de Lange syndrome (n=30)	Fragile X syndrome (n=28)	Prader-Willi syndrome (n=35)	Lowie syndrome (n=10)	Smith-Magenis syndrome (n=8)	Cri du chat syndrome (n=1)
Child age, range, mean (SD)	3-15 years 10.45 (3.34)	3-15 years 10.60 (3.89)	11-15 years 13.70 (1.26)	7-15 years 10.77 (2.20)	9-15 years 12.30 (2.45)	7-15 years 10.81 (3.18)	13 years (n/a)
Child Gender (n=males)	16	19	27	23	10	5	0
Mother's age range, mean (SD)	33-50 years 41.63 (5.15)	32-59 years 43.10 (6.58)	37-58 years 45.64 (6.05)	33-57 years 44.11 (6.53)	35-66 years 43.80 (8.74)	40-56 years 45.88 (5.48)	48 years (n/a)
Income- % below £25,000	9/33 27.3%	11/30 36.7%	14/26 53.8%	12/35 34.3%	3/10 30%	4/8 50%	0/1 0%
% Married or living together	28/33 84.8%	28/30 93.3%	19/28 67.8%	31/35 88.5%	9/10 90%	5/7 71.4%	1/1 100%

To measure maternal depression we used the depression sub-scale of the Hospital Anxiety and Depression Scales (HADS, Zigmond & Snaith, 1983, see Appendix 22). This scale contains seven four-point items, (e.g., “I feel as if I am slowed down” and “I still enjoy the things I used to enjoy”). The HADS has been widely used in community samples of parents of children with intellectual disabilities, and has excellent psychometric properties (e.g., Hastings, Beck, & Hill, 2005). Cronbach’s alpha for the present sample of mothers of children with rare genetic syndromes was .90.

Two measures of positive outcome were included in the current study: a short form of the 10 item Positive Affect Scale (PAS, Watson, Clark, & Tellegan, 1988), and the seven item Positive Gain Scale (Pit-ten Cate, 2003, see Appendix 12). A five item version of the PAS (the PAS5, see Appendix 23) was derived using the same procedure used to create the QRS-F7 (see above). Parents were asked to rate to what extent the five positive affect items had applied to them in the past week, on a scale ranging from “very slight or not at all” to “extremely.” The five items included were “Enthusiastic,” “Alert,” “Inspired,” “Determined,” and “Active”. Correlation of the PAS and the PAS5 was moderate to strong $r=.60$, $p=.53$. Internal consistency of the PAS5 within the current sample of mothers was good with a Cronbach’s alpha score of .88.

The Positive Gain Scale (MacDonald et al., in press; Pit-ten Cate, 2003,) assesses the direct positive aspects of having a child with a disability. Seven items including “Since having this child I feel I have grown as a person” and “Since having this child I feel I have become more determined to face up to challenges” are rated using a 5-point Likert scale from “0-strongly agree” to “4-strongly disagree”. The

lower the score, the higher the positive gain reported by parents. Cronbach's alpha for the present sample of mothers was .86.

Child measures

The Wessex Scale is designed to assess the social and physical characteristics of children with intellectual disabilities (Kushlick, Bludon, & Cox, 1973, see Appendix 24). It includes five subscales of mobility, self-help, continence, speech and literacy. There is evidence for good inter-rater reliability at subscale and item level (Kushlick et al., 1973), and the scales have recently been used successfully with populations of children with rare syndromes (Moss, Oliver, Arron, Burbridge, & Berg, 2009). The total self-help score was extracted for this study as a measure of the child's independence, which included three items: "Feed him/herself; Wash him/herself; Dress him/herself" which are rated from "1-not at all" to "3 -without help." Cronbach's alpha for these three items was .88.

The adaptive/social competence scale of the Nisonger Child Behavior Rating Form (NCBRF: Aman, Tasse, Rojahn, & Hammer, 1996, see Appendix 4) was used to assess children's positive behaviour including calm/compliant behaviours (e.g., followed rules) and adaptive/social behaviours (e.g., initiated positive interactions). The social competence scale includes ten items rated from "not true" to "completely or always true". The NCBRF has excellent psychometric properties (Aman et al., 1996), and Cronbach's alpha coefficient for the total adaptive/social competence score was .82 in the present study.

The Challenging Behaviour Questionnaire (CBQ; Hyman, Oliver, & Hall, 2002, see Appendix 25) was used to determine the frequency of child aggressive, self-injurious and destructive behaviour. For the purposes of the current study, three

items from this measure were combined to give a total challenging behaviour score. The items were (1) “Has the person shown self-injurious behaviour in the last month?” (2) “Has the person shown physical aggression in the last month?” and (3) “Has the person shown disruption and destruction of property in the last month?” An answer of ‘no’ was scored as 0, and ‘yes’ was scored as 1. These were summed to give a total score ranging from zero to three.

The Mood, Interest, and Pleasure Questionnaire (MIPQ: Ross & Oliver, 2003b; see Appendix 26) is a measure designed for use with individuals with intellectual disabilities and assesses overall psychological well-being and mood. The scale contains six items on mood (e.g., “in the last two weeks, did the person’s vocalisations sound distressed?”) and six items on interest and pleasure (e.g., “in the last two weeks, did the person seem to have been enjoying life?”). The higher the score, the greater the child’s overall mood, interest, and pleasure. It has been reported to have good test-retest (.87) and inter-rater reliability (.76) coefficients for the total score (Ross & Oliver, 2003b). Cronbach’s alpha for the total scale score was .82 in the current study.

The Health Questionnaire measures the presence and severity of 15 health problems (Oliver & Arron, 2008, see Appendix 27). Parents are asked to rate whether the child has been affected by the listed health problems in the last month (e.g., gastrointestinal difficulties, dental problems, skin problems). Items are rated from “0-no” to “3-severe”. Scores are summed to produce an Overall Health Score indicating severity of health problems for the previous month. For the current sample, Cronbach’s alpha was adequate at .69.

Procedure

The original data set contained 158 parents of children with rare syndromes, but we excluded any questionnaires that were completed by fathers ($n=9$) and also excluded any questionnaires that were completed by mothers whose children lived in residential care full-time ($n=4$), as we wished to determine the well-being of mothers who were regularly caring for their child at home.

All participants' contact details were on a central database of participants in previous studies held by the research team. The vast majority of parents on the database had originally been recruited via their child's syndrome support group, and had all agreed for their details to be held for contact about research projects. The questionnaires were mailed to the participant's homes, and the primary caregiver in the household was requested to complete the questionnaire. The completed questionnaires were then mailed back to the research team using the addressed envelopes provided in the questionnaire pack.

Results

The main analyses focused on regression models for the prediction of each of the five maternal well-being scores. Each model included all child variables as predictors alongside any demographic variables associated with the particular outcome. The demographic variables included in each analysis were selected because of their significant association with a maternal well-being measure (see Table 5.2). Table 5.2 also shows the correlations between each child variable and the maternal outcomes.

For each regression analysis, diagnostic statistics were examined to ensure that the statistical assumptions were met. Specifically, probability plots and histograms were examined to test for the assumption of the normality of distribution of regression

residuals. Residuals were found to be reasonably normally distributed in each of the five regression analyses.

Table 5.3 shows the summary of the results of the regression analyses for maternal distress variables (general maternal stress, rare syndrome stressors, and depression). Only three child variables emerged as statistically significant independent predictors of maternal distress. Child challenging behaviour was positively predictive of parenting stress, child self help was a negative predictor of parental stress; while child positive mood was a negative predictor of parental stress and depression. The effects of demographic variables were apparent for both the GSSS and depression scores. Mothers who were married or living with a partner reported fewer rare syndrome stressors and depression, and mothers of older children reported fewer stressors attributable to their child's rare syndrome. Only relatively small proportions of the total variance in maternal psychological distress scores were explained by the variables in this study.

Table 5.4 summarises the results of the regression analyses of maternal positive outcomes and shows a different pattern of child variable predictors. There were no significant predictors of maternal ratings of positive gain. However, positive affect was positively predicted by child positive mood. Additionally, mothers who reported having a higher annual income also reported higher positive affect. No other child or demographic variables significantly contributed to the variance in maternal positive outcomes. Once again, only small proportions of the variance in maternal positive outcomes were explained by the variables in the study.

Table 5.2. Correlations between demographic and child and maternal variables

Pearson's r	Child Age	Marital status	Annual Income	NCBRF adaptive/social	Wessex self-help	Mood, Interest, and Pleasure Questionnaire	Challenging Behaviour Questionnaire	Overall Health Score
QRS-F7 ¹	.007	.114	-.044	-.357**	-.292**	-.388**	.312**	.153
Depression	-.078	.185*	-.192	-.198	-.078	-.370**	.183*	.158
Genetic Syndromes Stressors Scale	-.209*	.213*	-.078	-.259**	-.236**	-.275**	.281**	.200
Positive Gain Scale	.027	.079	-.069	-.179*	.001	-.104	.174*	.100
Positive Affect Scale-5	-.052	-.162	.243**	.180*	-.008	.328**	.010	-.123

*p<.05

**p<.01

¹ Questionnaire on Resources and Stress- 7 item version

Table 5.3. Results of Multiple Regression analysis of maternal stress and depression

Regression Model/ Predictor variable	QRS-F7 ¹		Depression ²		GSSS ³	
	β	p	β	p	β	p
NCBRF Sociability/Adaptive	-.042	.140	.030	.750	-.092	.322
Wessex self help	-.167	.049	-.016	.860	-.126	.152
Child mood (MIPQ)	-.221	.012	-.308	.001	-.162	.067
Challenging behaviour (CBQ)	.183	.023	.063	.449	.121	.141
Overall Health Score	-.040	.615	.087	.931	-.016	.845
Marital Status	--	--	.189	.020	.265	.001
Child Age	--	--	--	--	-.191	.027

Table 5.4. Results of Multiple Regression analysis of maternal positive perceptions and affect

Regression Model/ Predictor variable	Positive Gain Scale ⁴		Positive Affect Scale ⁵	
	β	p	β	p
NCBRF Sociability/Adaptive	-.174	.088	.096	.323
Wessex self help	.106	.255	-.091	.305
Child Mood (MIPQ)	.016	.872	.281	.003
Challenging behaviour (CBQ)	.132	.136	.110	.192
Overall Health Score	.040	.650	-.005	.953
Annual Income	--	--	.173	.034

¹ R²=.220, F(5,136)=7.66, p<.001² R²=.154, F(6,136)=4.14, p<.01³ R²=.215, F(7,136)=5.14, p<.001⁴ R²=.052, F(5,136)=1.50, p=.193⁵ R²=.142, F(6,135)=3.71, p<.01

Discussion

The results in relation to the study predictions will be explored in turn. First, although child challenging behaviour was positively predictive of maternal stress, it did not emerge as the strongest predictor variable across maternal psychological distress measures. Second, consistent with our prediction, just one child variable was found to be a predictor of positive maternal outcomes. Third, a hitherto unexplored child variable (child mood) was predictive of maternal stress, depression, and positive affect. Finally, stressors related to the child's rare syndrome were predicted by the mother's marital status and the age of their child.

Although challenging behaviour was a predictive variable for general parenting stress, we had expected this to be a more consistent relationship with maternal psychological distress. A possible explanation for our lack of associations could be that the measure used in this study did not sufficiently differentiate the frequency of challenging behaviour between children. The measure also did not take into account the severity of the challenging behaviour and the subsequent wide range of frequency and severity of challenging behaviour likely to be among children in the same category may account for the weak predictive power. Additionally, the topographies of challenging behaviour were limited, and did not include stereotyped behaviour or verbal aggression. Defined measures of the frequency and severity of child challenging behaviour would be needed if this research was to be replicated.

Secondly, no child or demographic variables predicted mothers' positive perceptions of their children, and this is consistent with earlier research using multiple regression analysis on predictors of positive perceptions (Hastings et al., 2005). However, when looking at measures of mother's general positive affect, it was found that this was predicted by the ratings of child mood and by family

income. The overall trends suggest that 'positive' characteristics (child positive mood, higher income) contribute towards positive outcomes in parents. Therefore, although most positive and negative measures of maternal well being do appear to be independent, consistent with previous research (e.g., Blacher & Baker, 2006; Hastings & Taunt, 2002), child mood was predictive of both positive and negative outcomes. To our knowledge, this is the first study to show child variables which affect positive and negative maternal outcomes to a similar degree, and suggests that the two may not be entirely independent of each other under some circumstances. Therefore, further investigation is warranted into both of these findings to assess the independent variables underpinning both positive and negative outcomes and to evaluate the extent of independence or relatedness to the other.

The physical health of the child was not a significant predictor for any maternal outcome. A possible explanation is the sporadic nature of many health issues, which may lead to acute stress at the time (emergency hospital visits etc), may not be reflected in general measures of stress such as those used in the current study. Child self help only served as a negative predictor in one model; general parenting stress. Perhaps a more detailed measure on adaptive behaviour beyond self-help skills may be useful for future research. Adaptive behaviour not emerging as a strong predictor is not surprising given that in previous literature it has found to be associated with parental stress in some studies (Hodapp et al., 1997; Sarimski, 1997) but not others (Luesher et al., 1999).

Finally, a unique aspect of this study is that we measured stressors relevant to raising a child with a rare syndrome in a large sample of mothers who have children with various rare syndromes. No child variables were predictive of syndrome related stressors, but two demographic variables were significant predictors. Mothers who

were divorced or separated or had younger children were more likely to report higher stressors associated with their child's rare syndrome. Perhaps stressors resulting from a child's rare syndrome are more salient when the child is younger, when mothers may still be in the process of coming to terms with their child's diagnosis (Poehlmann et al., 2005). In the study detailed in Chapter 4, it was found that mothers of adults with rare syndromes did not feel that the rare syndrome of their child had much impact on their day-to-day lives, and the diagnosis of a rare syndrome was secondary to their offspring having intellectual disabilities. The stressors related to the rareness of the syndrome may have less influence as the child grows older, and further research is needed to extend the current findings.

One limitation of the current study is that the data are reliant on mothers own reports of their mental health, and their reports of the various attributes of their child. Therefore source variance may be a problem and information from wider sources (such as fathers or teachers), need to be considered to check the accuracy of maternal reports. Furthermore, no fathers were included in this study, and as studies have shown that mothers and fathers differ in their response to child variables (McCarthy et al., 2006), this study needs replicating with a sample of fathers. The data were also reliant on parental report of their child's diagnosis and so in future research efforts should be made to get clinical or genetic reports of the diagnosis.

Additionally, since the data are correlational, we are unable to infer causality. It is possible that child mood and interest affects parental stress, but it is also possible that this relationship is bidirectional over time (Hastings, 2002). Equally, there may be a third, unmeasured variable that influences both of these. This area would benefit from longitudinal research to determine how these variables influence each other over time.

Future research and practice

The present methodology could be extended to include other family members as part of the analysis, such as typically developing siblings. Hall, Burns, and Reiss (2007) found that both the behaviour problems of siblings and children with fragile X syndrome were equally associated with parental stress levels, and this could be further explored in future research. Family-centred approaches to research are important as families are dynamic, complex systems, and there is increasing evidence for multi-component conceptual models which incorporate variables such as other family members, demographic factors, and parental cognitions (Hall et al., 2007; Hastings et al., 2005; Lloyd & Hastings, 2008).

The findings from the present research suggest that mothers of children with rare genetic syndromes whose child has low mood are more likely to report higher levels of stress, depression, and lower positive affect. The research may help practitioners readily identify or anticipate parents at risk of high levels of stress given the characteristics of their child. This may be particularly salient for children with rare syndromes given the associated strong behavioural phenotypes. Also specific to the rareness of the child's syndrome is the finding that parents of younger children may report more syndrome-related stressors. Parents may benefit from support specific to issues arising from the rare genetic status of their child when their child is young when they may be assimilating to their child's diagnosis and also the specific needs of their child. Care providers could encourage links to syndrome support groups and provide links with professionals who are specialists in syndrome related issues (e.g., epilepsy consultants for parents of children with Angelman syndrome) in order to help parents anticipate and cope with any likely physical problems arising from their child's rare syndrome.

Chapter 6: General Discussion.

Parenting a child with an intellectual disability is a complex, conflicting experience, with feelings of joy and appreciation, sadness and guilt in an ever-changing flux. Differences in the behavioural phenotype of the child, such as problem behaviours, adaptive behaviours, and personality, are often linked to differences in family outcome (Briegel et al., 2008; Wulffaert et al., 2009). It is thus important to understand the behavioural phenotypes of rare genetic syndromes and how these influence the families of individuals with these syndromes. This thesis has attempted to expand existing knowledge of the experiences of parents of children with rare genetic syndromes. First, a review of the existing literature on parents and siblings of children with rare syndromes was conducted, with suggestions for possible directions for future research. Most striking about the reviewed literature was the diversity of approaches taken by researchers to examine parents of children with rare syndromes; as a result, studies were often exploratory in nature and rarely actively built on preceding literature. A stronger focus, using clearer methodological approaches or existing models of parental stress (e.g., Hastings, 2002; McCubbin & Patterson, 1983) is recommended.

In Chapter 2, a matched group approach was used to analyse child behaviour and parental adjustment in children with either Down syndrome, autism, or mixed aetiology intellectual disability. Despite matching the groups, some differences in child behaviour emerged (e.g., children with Down syndrome were rated as more sociable, and children with autism had more behaviour problems). Very few differences in maternal stress, anxiety, depression, and positive perceptions were observed between the three groups. Therefore, there was little evidence of a Down syndrome advantage (Hodapp et al., 2003). This is similar to previous research that controlled for child or socio-economic variables (Cahill & Glidden, 1996; Stoneman,

2007). This study highlighted the need for closely matched groups where possible in order to reduce threats to internal validity when exploring parental distress and well-being.

Chapter 3 of this thesis was concerned with parents of children with either Angelman, Cri du Chat, or Cornelia de Lange syndrome, whose children engaged in high frequencies of challenging behaviour. That is, the groups were designed to be reasonably well matched for challenging behaviour at the outset. A group of parents of children with autism were included as a 'benchmark' group for parental distress. Parents of children with Angelman and Cri du Chat syndrome were more likely to report clinical levels of anxiety and depression symptoms than normative samples. The general pattern of results revealed that parents of children with Angelman syndrome reported the highest scores on negative outcomes, parents of children with Cornelia de Lange syndrome the lowest, with parents of children with Cri du Chat syndrome and autism being in between the two groups. This is the first study to use parents of children of autism as a 'benchmark' comparison group, and demonstrates the importance of using parents of children with well-researched conditions when trying to establish how parents of children with relatively under researched rare syndromes are coping. Additionally, this is the first study to quantitatively measure positive well-being and perceptions of positive gain in parents of children with rare syndromes, and demonstrates the importance of including positive measures in future research in order to gain a well balanced and realistic view of family adjustment.

The research described in Chapter 4 gives an in-depth insight into the experiences of mothers of adult offspring with Angelman, Cri du Chat, and Cornelia de Lange syndrome, focusing particularly on their experiences of receiving social

and medical services for their offspring. This qualitative study showed that parental involvement with their offspring continues into adulthood, and in particular, issues surrounding obtaining and retaining adequate social and medical support places significant strain on mothers. This is similar to previous findings with mothers of other adults with intellectual disabilities (Todd & Jones, 2005; Todd & Shearn, 1996; Shearn & Todd, 1997).

The research in Chapter 4 is the first of its kind to explicitly investigate whether the *rareness* of their offspring's syndrome had any impact upon mothers. It was found that it made little difference to how mothers perceived their offspring or their relationship with social care services, but did create unique difficulties in two areas: (1) In encounters with medical professionals who knew little about their offspring's rare syndrome, and (2) Not knowing how the rare genetic syndrome may affect their offspring during their adult life. This study highlights the struggles that some mothers encounter with social care and medical services, and the large impact inadequate services have on the wider family. The findings could be used to inform possible areas of inquiry among larger scale studies among parents of children with rare syndromes.

The final empirical study (Chapter 5), adds to the literature as it is the first study to look at predictive child and demographic variables in negative and positive measures among a large mixed sample of mothers of children with various rare syndromes. It was found that a child measure unused before in family adjustment literature (the MIPQ: Ross & Oliver, 2003a) was predictive of both negative and positive maternal measures; whereas previous research has found negative and positive outcomes to be independent of each other (Blacher & Baker, 2006; Hastings & Taunt, 2002). Additionally, mothers of children in Chapter 5 reported higher

syndrome-related stress when their child was younger, which may be related to the finding in Chapter 4 in which mothers of adults with rare syndromes felt that the rare syndrome was secondary to the fact they have an intellectual disability. This trend may suggest that stressors related to the rareness of the child's syndrome decrease as the child grows older. The findings show that a wide range of child variables need to be taken into account when investigating adjustment among parents of children with rare syndromes, and suggest that stressors related to the rareness of their offspring's syndrome becomes less salient as the child grows older.

Theoretical implications

The findings from the four empirical studies in this thesis suggest that conceptual models used in the wider family functioning research on children with intellectual disabilities are also likely to be applicable to families of children with rare genetic syndromes. Many of the findings are not unique to families of children with rare syndromes (such as lower child adaptive behaviour and higher levels of challenging behaviour being associated with higher parental stress). Therefore, models used for families of children with intellectual disabilities, such as the Double ABCX model (McCubbin & Patterson, 1983), Lazarus and Folkman's (1984) Process model of Stress and Coping, and the Family Adjustment and Adaption Response model (FAAR: Patterson, 1983) are all likely to be applicable to families of children with rare syndromes. The current findings also suggest that there are likely to be some variables that uniquely influence parents of children with rare syndromes that could be added to any conceptual model on functioning.

Overall, similar to existing models of family adaption, a conceptual model of families of children with rare syndromes would incorporate socio-demographic factors (such as income, marital status), as overarching circumstances such as low

socioeconomic status and not having a partner may contribute to parents reporting higher levels of stress and depression (as found in Chapter 5). These circumstances may place further strain on parents who may be already dealing with the stressors of raising a child with an intellectual disability.

There is increasing interest in the positive aspects that children with intellectual disabilities bring to their parents and family (Blacher & Baker, 2006; Hastings & Taunt, 2002). Positive parental adaptation has rarely been looked at in prior research on parents of children with rare genetic syndromes. The findings in Chapters 3 and 5 demonstrate that positive measures are salient to parents.

Therefore, similar to the wider family adjustment research, it is important to include measures of positive adjustment in conceptual models of parenting in order to maintain a realistic view of parenting.

Another aspect of family functioning explored by the current thesis was the impact of the demands that obtaining and maintaining support for their offspring from social support services affected mothers (Chapter 4). These accounts reflect personal experience and mothers expressed how dealing with persistent issues arising from social care provision could lead to increased stress. The relationship with or support received from statutory services has not, to the authors knowledge, been explicitly included within any family adjustment models, although given previous research is, again, unlikely to be unique to parents of adults with rare syndromes (e.g. Shearn & Todd, 1997). These findings suggest that this aspect of caring for an individual with rare genetic syndromes can place considerable, long term strain on mothers, and may be an important variable to consider both in conceptual models and in future research among parents of both adults and children with rare genetic syndromes and in the wider family literature.

All the above factors are interrelated and most are likely to be bi-directional. For example, Hastings (2002) proposed that the relationship between parental stress and child behaviour problems is bidirectional, as challenging behaviour is associated with higher parenting stress this in turn may affect parental behaviour and how parents directly react to the challenging behaviour of their child.

The behavioural phenotype, adaptive and challenging behaviour of the child, family resources, and the outcome variables of parental mental health are taken into account in general family models, and are also applicable to families of children with rare syndromes. Therefore, there appear to be no substantial differences in the fundamental family functioning components between families of children with rare genetic syndromes and other families of children with intellectual disabilities. However, there is evidence in this thesis that there may be some additional unique aspects to parenting a child with a rare genetic syndrome to consider: (1) The function of challenging behaviour of the child, and (2) Stressors associated with the rareness of a child's syndrome.

Firstly, there is a possibility that the functional profile of the challenging behaviour of the child may also influence parental adjustment. In Chapter 3, both mothers and fathers of children with Angelman syndrome consistently reported higher stress, anxiety and depression levels than parents of children with Cri du Chat syndrome, Cornelia de Lange syndrome, and autism, although all children displayed similarly high levels of challenging behaviour. It may be that the behavioural phenotype of Angelman syndrome, which is associated with high levels of attention-seeking behaviours (Tunicliffe, 2009), may contribute to increased parental stress. Therefore, the function of a given challenging behaviour may be a useful additional component in a conceptual model of families of children with rare syndromes.

Secondly, the findings in this thesis also suggest that issues pertaining to the rareness of a child's syndrome are salient to parents, and unique stressors may arise from this (e.g., not having access to professionals who have knowledge about the syndrome), which need to be incorporated into any conceptual model. Additionally, there is evidence that a different set of dynamics may emerge as offspring with a rare syndrome age. In particular, there is evidence presented in Chapter 5 which suggest that parents do not rate stressors related to the rareness of their child's syndrome as highly as the child gets older. Further longitudinal research is needed to explore these results.

These chapters, taken together, demonstrate both the similarities and differences between families of children with intellectual disabilities and those of children with rare genetic syndromes. They help form an account of additional variables and concepts which need to be considered when exploring adjustment among parents of children with rare syndromes. The perspectives outlined here will be expanded and further theoretical and research implications will be discussed.

To date, there are no consistent theoretical basis from which parents of children with rare genetic syndromes have been investigated. One approach which may be useful is the concept of indirect effects, in which behaviours characteristic of a given syndrome elicit different reactions from people in their environment than the behaviours characteristic of another syndrome (Hodapp, 1997). This may be a useful approach when exploring the affect the function of a child's behaviour has on parents. Although some work on indirect effects has been conducted with parents of children with Down syndrome, this concept has not been directly examined among children with rare syndromes.

Fidler (2003) found that parents of individuals with Down syndrome (aged 5-20) raised their voice pitch and had a slightly wider pitch variance than parents of individuals with other forms of intellectual disability, regardless of the chronological age of the individual with Down syndrome. These voice characteristics are similar to those made towards younger children. Therefore individuals with Down syndrome elicit a type of parental behaviour more often used with much younger children, this finding links in to the concept of indirect effects (Hodapp, 1997). Much work remains to be conducted on discovering the most powerful eliciting characteristics of the behavioural phenotype of children with rare syndromes, and why these are difficult (or advantageous) for parents. Perhaps future work on aggression among children with Angelman syndrome (which is often motivated by desire for attention; Tunicliffe, 2009) and how this affects parents may give further evidence for the concept of indirect effects.

The examination of parental cognitions such as attribution theory would be interesting to apply to research with parents of children with rare syndromes (Bugental, Johnston, New, & Silvester, 1998). Attribution theory, as applied to family adjustment, is concerned with the way parents perceive (or attribute) the behaviour of their children. As parents of children with rare genetic syndromes will be aware that there may be a definite biological basis for some of their child's behaviour problems, will parents be more likely to attribute challenging behaviours to factors beyond their child's control (external attribution) than within the child's control (internal attribution)? If this is the case, how does it impact the wellbeing of parents and their subsequent interactions with their child? Parents who attribute challenging behaviour to factors beyond the child's control may diminish feelings of responsibility for the problem (Himelstein, Graham, & Weiner, 1991). One study

found that parents who attributed their child's challenging behaviour to physical causes are less likely (than parents who do not make this attribution) to feel that behavioural interventions which focus on environmental variables will be effective (Reimers, Wacker, Derby, & Cooper, 1995). This is important to investigate among parents of children with rare genetic syndromes, as if a parent attributes a child's challenging behaviour as an inevitable part of their child's syndrome, it may be that they will be less likely to rate environmental-based behavioural interventions; which could in turn lead to lower adherence to a behaviour program.

Lastly, as well as focussing closely on particular parental cognitions or the indirect effects of a syndrome; the wider family context needs to be taken into account. The findings from the literature review and the empirical studies in this thesis demonstrate the need for a clearer focus on conceptual models of family adjustment. Although there is evidence of a strong relationship between higher levels of maladaptive behaviour and increased maternal stress in parents of children with rare syndromes (Briegel et al., 2008; Hodapp et al., 1997; Richman et al., in press; Wulffaert et al., 2009), we can not assume that this is due to child behaviour affecting parental stress levels (Hastings, 2002; Lewis et al., 2006). The relationship may not be unidirectional, but circular, with maladaptive behaviour leading to higher maternal stress which in turn leads to mothers perceiving maladaptive behaviour to be worse than it actually is (Hastings, 2002). In order to further understand these complex processes, research is needed from a family systems perspective, and the Double ABCX model (McCubbin & Patterson, 1983), and this will be discussed further in the following section.

Research implications

Due to the lack of a cohesive literature investigating parents of children with rare syndromes, it was necessary for some of the empirical work in this thesis to be exploratory. The strengths of the research in this thesis lie in the unique contribution to family adjustment research, in part due to the methodological approaches taken. This section describes why these approaches need to be considered in future research, and how they could be extended; (1) The use of closely matched groups, (2) The use of well-established aetiologies as comparison groups for parents of children with rare syndromes, (3) The inclusion of positive measures in family adjustment research, (4) The use of qualitative methods for in-depth insights into how parents perceive themselves and the demands they are under, (5) The importance of including a wide range of child and demographic variables in quantitative research, and (6) Investigating stressors that are associated with the *rareness* of their child's syndrome.

Firstly, when comparing groups of parents according to the aetiology of their child, there is a need to use matched groups of children with rare syndromes to control for potentially confounding variables, which will reduce the likelihood of internal validity being compromised. On a practical level, this may be difficult to achieve as a given genetic syndrome may be so rare that achieving closely matched groups is impossible. Unless large matched samples become achievable (perhaps through international collaboration) the field may continue to be reliant on statistical techniques which help control for potentially confounding child and demographic variables.

Secondly, this is the first study to use parents of children of autism as a 'benchmark' comparison group alongside parents of children with rare syndromes. This is more informative than comparing two groups of parents of children with

different rare genetic syndromes when little is known about either group. This helps gain an immediate understanding of the levels of stress reported by a particular group of parents. The use of a benchmark group such as autism could allow researchers to quickly identify and target potential at risk groups who report very high levels of stress.

The studies in this thesis are the first to quantitatively measure positive well-being and perceptions of positive gain in parents of children with rare syndromes. It is also the first study to find that positive child mood was a predictor of positive outcomes for parents of children with rare syndromes. The results are encouraging and demonstrate the importance of continuing to include positive measures in future research to achieve a well balanced view of family adjustment. The below quote (from an interview conducted for the DVD-ROM) from a mother of a 12-year old boy with Cornelia de Lange syndrome demonstrates the reality of sadness intertwining with a great deal of joy in everyday family life, and why it is important to continue to strive to reflect this accurately in the research literature.

He's a really happy, most of the time placid little boy, who's enriched mine and my husbands life immeasurably since we've had him (...) He's a lot more part of our lives in a way then perhaps a normal child would be cos you have to do so much for him for so much longer then you would for a normal child.

Also there's a lot of sadness in a way because you, you miss out on a lot of things that if you've got an ordinary child you sort of just take for granted.

You know he never comes home and tells us what he's done at school he's not into anything like sport or mad keen on football or anything like, so you miss a lot of that, and you have to take a lot of, you take pleasure in things that are a

lot more minor, I certainly take pleasure from him in more minor things than you do the bigger things.

Fourthly, in future research on families with children with a rare genetic syndrome, perhaps a flexible, mixed research approach combining both qualitative and quantitative methods may be useful. As much of the research on parents of children with rare syndromes is still in its infancy, a ‘bottom up’ approach to the data, in which individual affects of the syndrome on parental well-being are looked for rather than the general affects, may be useful. Qualitative research methods are likely to result in richer data and reveal hitherto unknown aspects of a child’s behavioural phenotype which parents find particularly distressing. This information is valuable in itself, but could also be used to inform larger quantitative studies, and could potentially add to the literature surrounding the indirect effects of a child with a rare genetic syndrome in the family (Hodapp, 1997).

Most previous research has concentrated on the child variables of challenging and adaptive behaviours. The research in Chapter 5 demonstrates the importance of including a wide range of other child variables that may also related to parental measures. The exclusion of potentially salient child variables may result in an inaccurate portrayal of family adjustment. This could also include unusual, syndrome-specific behaviours in order to assess the impact of these on parental well-being. Unusual behaviours associated with a specific syndrome were only investigated in two studies, both of which found a strong association with parental stress (Hodapp et al., 1997; Laurvick et al., 2006). Further research is warranted on the identification of unusual features of some behavioural phenotypes and how these affect parental well-being.

Finally, measures related to the rareness of the child's syndrome are often overlooked in research. The study detailed in Chapter 5 suggest that mothers report more syndrome-related stressors when their child is younger. This finding, taken together with mothers of adults not placing particular importance on the rareness of their offspring's syndrome suggest that stress related to the rareness of a syndrome may reduce as the child gets older. The below quotes demonstrate the contrast between mothers of infants and mothers of adults with a rare syndrome. The below quote is from an interview (conducted for the DVD-ROM) with a mother of a three year old boy with Cornelia de Lange syndrome; this gives an insight into the complexities of the day-to-day tasks involved in the first years of having a child with a rare syndrome.

When my son was around 1 year old, there was a constant trickle of phone calls, letters, and appointments. He's got about a dozen consultants he sees on a regular basis to do with different aspects of his syndrome and development. I wasn't prepared for that constant dripping tap over months and months of more and more people getting involved with him and that was really hard work (...) So I felt under pressure because I wanted to do the right thing by him, but equally trying to get all those little bits in the day, like a bit of physiotherapy, a bit of trying to sort out his diet and sort out things to do with his textures, because he is very tactile defensive. That was just immense.

When this is contrasted to a mother of an adult with a rare syndrome (Grace; participant in the study in Chapter 4) we see that the rareness of her son's syndrome does not affect any aspect of her current caregiving role.

He has a learning disability and therefore you know we just live with that, whether it's Cri du Chat or any of the others, really at this stage in his life, it might have made a difference earlier on.

It is important to continue to investigate this unique experience of parents of children with rare syndromes, as findings may have implications for clinical interventions. The development of the GSSS has gone some way to addressing this issue, but much more work is needed to explore this unique aspect of families with children with rare syndromes. The use of the GSSS in future research may be a useful starting point. Additionally, qualitative analysis using grounded theory (Willig, 2001) may be helpful to ask parents directly about how they think the rareness of their child's syndrome affects their lives and those of their family. Qualitative interviews could be repeated over time from time of genetic diagnosis onwards to chart how families change as their child grows older.

Further research developments

As well as the methodological points above which were explored in the current thesis, there are other potential research developments which were not explored in this thesis directly but could help expand the research area on the adjustment of families with a child with a rare syndrome. One particular area of development from the work in this thesis could be on parents of children with Angelman syndrome, who reported strikingly elevated levels of stress, anxiety and depression, even when compared to parents of other rare syndromes and parents of children with autism (Chapter 3). The behavioural phenotype of children with Angelman syndrome could be examined to help determine why this might be. An interesting area to investigate would be the heightened sociability of children with Angelman syndrome, and the effect of this on parents. Anecdotal reports from

parents indicate that children have a strong and constant desire for social attention which can be very demanding (one mother reported she could not go to the bathroom without her child following her and wanting attention). There is also evidence that challenging behaviour in children with Angelman syndrome may arise from a desire for social attention (Tunnicliffe, 2009). Thus, this heightened sociability aspect of children with Angelman syndrome may contribute to parental stress, and the concept of indirect effects (Hodapp, 1997) may be a useful approach with which to address this issue.

Another possible expansion of the work in this thesis is to collect longitudinal data to investigate how parents of children with rare syndromes adjust and adapt over time. Although some cross-sectional studies on families of children with rare syndromes have been published using samples with a wide age range - from 1 year old to 46 years old (Wulffeart et al., 2009); collapsing the sample across this wide age range does little to inform of any changes attributable to supporting an individual at different stages of the life cycle. Longitudinal research has been used with parents of children with Down syndrome (Hauser-Cram, Warfield, Shonkoff, & Krauss, 2001; Most et al., 2006), and could be extended to work with parents of children with rare genetic syndromes. It may be useful to follow families over the years concentrating on salient developmental periods such as infancy, middle childhood, adolescence, and young and mid-adulthood. Some researchers have argued that the optimal design for studying behavioural phenotypes is to combine initial cross sectional designs with longitudinal follow-up (Karmiloff-Smith, Scaife, & Thomas, 2002); A longitudinal approach may offer useful insights into the question of how parents perceive stressors related to their child's rare syndrome, as

well as whether the developmental trajectories of behavioural phenotypes affect parents.

Future research would also need to consider members of the family other than parents. The family systems perspective proposes that all members of a family are interrelated, so something that affects one family member also affects all individual members of that family, as well as the family system as a whole (Minuchin, 1985). Thus, family members other than parents should be taken into account. There is evidence that siblings of children with intellectual disabilities/autism can be negatively affected by the child with a disability (Hastings, 2003; Roderigue, Geffen, & Morgan, 1993). Siblings can be affected directly (i.e., have to accommodate their brother or sisters additional needs on a day-to-day basis) and indirectly (i.e. if parents are undergoing stress this may affect family functioning as a whole). Little research has been conducted on siblings of children with rare syndromes, but this is a worthy area of future research.

Typically developing siblings also need to be considered as part of the family system, as by excluding data from siblings (as independent variables) researchers may be missing potential sources of family stress. Hall et al., (2007) found that sibling problem behaviour was just as influential on parental stress as the behaviour problems of their brother or sister with fragile X syndrome, and so need to be included when investigating the family system.

Another neglected issue in previous research on rare syndromes, and one that has not been addressed in the current thesis, is that only parent and sibling reports of family adjustment have been considered. What do people with mild intellectual disabilities and rare syndromes say about their families and what they mean to them? Nothing is

known about how individuals with rare syndromes feel about their families, although it could be a potentially insightful area of inquiry.

Methodological limitations of the current research

While the research in this thesis makes several unique contributions to the literature surrounding family adjustment for parents with children with rare syndromes, there are some limitations that need to be borne in mind.

The largest methodological limitation we encountered was the difficulty in recruiting large numbers of parents of children with the same rare genetic syndrome. The consequential small sample sizes of the parental groups have implications in two areas; first, the small sample sizes limited the type of statistical analysis we could perform on the data. More complex analyses were therefore not feasible in the study described in Chapter 3 due to the high likelihood of Type 1 error. Secondly, we do not know to what extent these results can be generalised to the wider population. Small sample sizes are an inherent problem when examining rare syndrome populations and large-scale, international studies may be needed to recruit larger sample sizes.

Another sampling issue inherent in this thesis is that every parent in the studies Chapters 3, 4, and 5 were recruited via their child's national syndrome support group. There are many possible reasons why parents may join a support group, they may or may not be more distressed, more pro-active, or have children with more severe disabilities than parents who do not join a support group. It is not known whether the characteristics of parents who join support groups differ from those that do not, and therefore it is difficult to evaluate the representativeness of parents who are members of syndrome support groups (Horsler & Oliver, 2006).

Researchers must continue to be aware of the implications of this sampling method, and future efforts should be made to recruit participants through other means, such as via General Practitioners, or special education settings.

A broader issue is that existing questionnaires and other instruments often do not support research on children with rare genetic syndromes, as they are not designed for use with individuals with severe/profound levels of intellectual disability. Instruments measuring adaptive behaviour such as the VABS-II (Sparrow et al., 2005) are not ideal for use with children with rare syndromes as they were originally developed for typically developing individuals. Norms from the VABS-II (Sparrow et al., 2005) do not extend in sufficient detail to the lower levels of ability associated with many rare genetic syndromes. Therefore, there is likely to be a range of ability among children in the lower end of the adaptive scale which is being missed.

An example of the problems caused by using the VABS-II (Sparrow et al., 2005) in the present thesis was that children who communicated using sign language were classed as not having any communication beyond simple gestures (e.g., pointing), although this was not true. One of the participants with Cri du Chat syndrome used sign language to communicate complex concepts such as “Sounds like rain” but - in accordance with the VABS-II criteria - had to be classed as having no communication beyond gesturing at objects, as she did not use speech to communicate. This highlights the need to develop and use measures of adaptive behaviour which reflect these differences and thus are suitable for use with individuals with severe/profound intellectual disability.

Finally, the data in all four empirical studies in this thesis were reliant on self-report data from parents, and so caution must be used when interpreting these

results. Stone et al., (1998) compared ongoing, frequent reports on a palm-top computer with retrospective reports on coping and found that behavioural coping methods were over-represented, and cognitive coping methods were under-represented in retrospective reports. These findings are likely to be extended to include self-report about parental well-being. A way of overcoming these issues is to use multiple informants. Child behaviour problems for example, could be rated by teachers or another immediate family member to get validation of parental report of behaviour problems. It may also be possible to collect data on problem behaviours by direct observation of parent/child interactions. This approach may give further insight into the nature of these relationships and how they relate to parental self-report measures of stress and coping. This approach has been used successfully with parents of children with Down syndrome (Atkinson et al., 1995) and could be extended to children with rare syndromes

Practical implications

Although there has been a recent push to take type of syndrome and associated cognitive strengths and weaknesses into account in educational and clinical settings, (Hodapp & Fidler, 1999; Hodapp & Dykens, 2001), there is no discernable movement in the literature towards how best to support families of children with rare genetic syndromes. The following potential applications of the research will be discussed (1) Coping with the rareness of their child's syndrome, (2) Management of challenging behaviour, and (3) Directly targeting parental stress and cognitions.

Coping with the rareness of their child's syndrome.

In terms of supporting parents with the issue of the rareness of their child's syndrome, two main approaches emerge from the research in this thesis. Firstly,

continued research on defining and refining the behavioural phenotype across the entire lifespan of individuals with rare genetic syndromes is important. Although the focus of this review was on current family functioning in parents of children with rare syndromes, many parents report that worrying about their child's future is an additional stressor (data from the Genetic Syndrome Stressors Scale, Chapters 3 and 5). We also found that parents of adults with rare genetic syndromes found the lack of available information about their offspring's likely lifespan or potential aging issues disconcerting and stressful (Chapter 4) although overall the rare syndrome diagnosis did not affect their day-to-day lives. Continuing to study the behavioural phenotype in individuals with rare syndromes into adulthood and making this information widely available may help reduce parental anxiety, and help parents prepare for any developmental aging issues which may be associated with the syndrome. Secondly, the evidence suggesting that parents of younger children report greater levels of stress relating to the rareness of their child's syndrome (Chapter 5) has implications for intervention and some parents may benefit from greater support for any issues arising from the rareness of their child's syndrome in the first few years of their child's life. Care providers could encourage parents to join to support groups, which may also be beneficial. One mother described the relief she felt after meeting other parents at a support group conference.

The support group conference made a massive difference because then I felt I wasn't the only person in the world, and then I got scared then because I didn't want to leave the conference as I felt protected and in a world where everybody knew what was happening and everybody understood.

(Mother of 3 year old boy with Cornelia de Lange syndrome; excerpt from interview for the DVD-ROM)

Additionally, parents could also be directly supported throughout their child's early years by care providers giving information about the syndrome and ensuring links are made with appropriate professionals for any issues which are associated with their child's syndrome (e.g., a gastrointestinal specialist for parents of children with Cornelia de Lange syndrome).

Behaviour management.

Interventions which directly target behaviour problems in children with rare syndromes may also help reduce parental stress. Currently, some researchers are investigating whether challenging behaviors in children with rare genetic syndromes are also influenced by social/environmental variables. For example, there is evidence of social reinforcement playing a role in the maintenance of self-injurious behaviour in children with Cornelia de Lange syndrome (Moss et al, 2005), and children with Angelman syndrome show high levels of attention-maintained and demand-escape challenging behaviours (Tunnicliffe, 2009).

Evaluation of the influence of environmental variables in the maintenance of self-injury and aggression within and across syndromes has clear implications for behavioural interventions, as if a specific mode of reinforcement is characteristic of a syndrome, this information can inform assessment and intervention. The evidence based learning theory approach to problem behaviour used for children with autism (Anderson & Romanczyk, 1999; Harris & Delmolino, 2002) is likely to be applicable to children with rare syndromes but this is, as yet, unevaluated. It is important to establish whether this approach is useful and how it should be modified to take account of syndrome specific characteristics. This will help practitioners and parents accurately target any problem behaviours from a young age, and given the relation between problem behaviour and parental stress; targeted, effective

behavioural interventions may improve parental well-being (Hudson & Gavidia-Payne, 2002). Any behaviour interventions may also need to include interventions looking at parental attributions of their child's behaviour, or overall parental stress. This is particularly important as parents experiencing significant stress may be less effective at carrying out behavioural intervention programs (Hastings & Beck, 2004).

Parental stress and cognitions.

Interventions could be primarily focused on reducing parental stress directly. As the research on behavioural phenotypes and family adjustment grows, care providers may be able to anticipate family stress given the behavioural phenotype of the syndrome. Additionally, interventions could focus on parental cognitions which have been shown to be related to parental stress. Some studies suggest that parental burden is directly related to coping strategies and family functioning (Luescher et al., 1999). Thus, interventions teaching parents useful coping strategies may alleviate parental stress. More recently, interventions aimed at increasing parental acceptance (i.e., accepting their child as a person in their own right) are gaining credence, and although the majority of research surrounding acceptance is on parents of children with autism (e.g., Singh et al., 2006), it may also be applicable to parents of children with rare genetic syndromes. There are many ways in which parents of rare syndromes can be supported, and the best approach may be a flexible, holistic approach that incorporates all of the three areas above.

Dissemination

In terms of practical outcomes, the Three Syndromes project has resulted in the production of three informational DVD-ROM's; one each on Angelman, Cornelia de Lange, and Cri du Chat syndromes (see Appendix 28 for examples of

content and pictures from the DVD-ROM about Angelman syndrome). The DVD-ROM's were written by the Three Syndromes research team and were designed to help give information to professionals or family members who wish to know more about a syndrome. It includes information on genetics, parents recalling how they coped when their child was diagnosed, information on health issues associated with the syndrome, and on adaptive and challenging behaviour. The DVD-ROM's have been distributed to the international parent support groups to help highlight the research and to help parents to manage their child's challenging behaviour. This DVD-ROM is an example of one way in which researchers can disseminate information in an accessible way to parents and professionals.

Conclusions

In conclusion, the present thesis demonstrates the high levels of parenting stress, anxiety, and depression undergone by many parents of children with rare genetic syndromes, and the child variables that may contribute to the stress levels of parents. In addition, positive adjustment and perceptions have been demonstrated to be salient to parents of children with rare syndromes. This thesis has highlighted that parental involvement continues into their offspring's adult years, where different stressors become relevant. This thesis has added to the field of psychological adjustment in parents of children with rare genetic syndromes by highlighting suitable methodologies for use in future research, and potential applications of the research.

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Appendices

Appendix 1

Demographic Questionnaire – Special Needs and Families Research Project

The following questions ask for background information about you, your child with special needs, and your family. Please tick the appropriate boxes or write in the spaces provided.

1. Are you male or female? Male Female

2. What was your age in years on your last birthday? _____

3. What is your current marital status?

Married, and living with spouse.....

Living with partner.....

Divorced/Separated/Single and NOT living with a partner.....

4. In total how many people currently live in your house? _____ Adults _____ Children

If there are other children living in the house how are they related to your child with special needs (e.g. biological brother, step brother) and how old are they? – *Please list ALL children*

.....

5. Please tick the boxes next to all of the educational qualifications that you hold

No formal educational qualifications.....

GCSE, CSE, GCE, O Levels or equivalent.....

GCE, A Levels, HNC, GNVQ or equivalent.....

HND, other Diploma, or equivalent.....

Polytechnic/University ordinary or honours degree.....

Masters or Doctoral degree.....

10. What is your relationship to your child with special needs (e.g., mother, father, stepmother, grandmother, adoptive parent)? _____

11. How old is your child with special needs? _____ years _____ months

12. Is your child with special needs male or female? Male Female

13. Please tick the boxes below to indicate any diagnoses/conditions that apply to your child with special needs

Learning Disability (“Intellectual Disability”).....

Autism.....

Cerebral Palsy.....

Down Syndrome.....

Other syndrome (please specify) _____

14. Does your child with special needs have sensory impairment that interferes with his/her day to day life?

Yes No

If yes, what is this impairment?

Visual impairment? Yes

Hearing impairment? Yes

15. Does your child with special needs currently suffer from epileptic fits? Yes No

16. Does your child with special needs have problems with mobility that mean it is difficult for them to move around independently (e.g. needs to use a wheelchair)? Yes No

17. Does your child with special needs have any other health problems not already mentioned? Yes No

If yes, then please specify _____

19. Recent data from research with families of children with special needs has shown that a family's financial resources are important in understanding family member's views and experiences. With this in mind, we would be very grateful if you could answer the additional question below. We are not interested in exactly what your family income is, but we would like to be able to look at whether those with high versus lower levels of financial resources have different experiences.

What is your current total annual family income? Please include a rough estimate of total salaries and other income (including benefits) before tax and national insurance/pensions.

Please tick one box only:

- Less than £15,000.....
- £15,001 to £25,000.....
- £25,001 to £35,000.....
- £35,001 to £50,000.....
- £50,001 to £75,000.....
- £75,001 to £100,000.....
- £101,001 to £150,000.....
- £151,001 to £200,000.....
- £201,001 or more.....

Appendix 2

Behaviour Problems Inventory

On the following pages you will find generic definitions followed by specific descriptions of three types of behavior problems: self-injurious behaviors (items 1-15), stereotyped behaviors (items 16-40), and aggressive/destructive behaviors (items 41-52).

Please indicate which behaviors you have observed in your child with special needs during the past two months by circling the number in the appropriate boxes to indicate (a) how often the described behavior typically occurs (frequency) and (b) how much of a problem the behavior represents. If the behavior has never been observed during the last two months, circle the number "0".

SELF-INJURIOUS BEHAVIOR

Generic definition: *Self-injurious behavior (SIB) causes damage to the person's own body; i.e., damage has either already occurred, or it must be expected if the behavior remained untreated. SIBs occur repeatedly in the same way over and over again, and they are characteristic for that person.*

	Never	Frequency				Degree of Problem		
		monthly	weekly	daily	hourly	slight	moderate	severe
1. Self-biting (so hard that a tooth print can be seen for some time; bloodshot or breaking of skin may occur)	0	1	2	3	4	1	2	3
2. Hitting head with hand or other body part (e.g., face slapping, knee against forehead) or with/against objects (e.g., slamming against a wall, knocking head with a toy)	0	1	2	3	4	1	2	3
3. Hitting body (except for the head) with own hand or with any other body part (e.g., kicking self, slapping arms or thighs), or with/against objects (e.g., hitting legs with a stick, boxing the wall)	0	1	2	3	4	1	2	3
4. Self-scratching (so hard that reddening of the skin becomes visible; breaking of the skin may also occur)	0	1	2	3	4	1	2	3
5. Vomiting and rumination (deliberate regurgitation of swallowed food with rumination)	0	1	2	3	4	1	2	3

6. Self-pinching (so hard that reddening of the skin becomes visible; breaking of the skin may occur)	0	1	2	3	4	1	2	3
7. Pica: Mouthing or swallowing of objects which should not be mouthed or swallowed for health or hygiene reasons (non-food items such as feces, grass, paper, garbage, hair)	0	1	2	3	4	1	2	3
8. Stuffing objects in body openings (in nose, ears, or anus, etc.)	0	1	2	3	4	1	2	3
9. Pulling finger or toe nails	0	1	2	3	4	1	2	3
10. Stuffing fingers in body openings (e.g., eye poking, finger in anus)	0	1	2	3	4	1	2	3
11. Air swallowing resulting in extended abdomen	0	1	2	3	4	1	2	3
12. Hair pulling (tearing out patches of hair)	0	1	2	3	4	1	2	3
13. Extreme drinking (e.g., more than 3 liters per day)	0	1	2	3	4	1	2	3
14. Teeth grinding (evidence of ground teeth)	0	1	2	3	4	1	2	3
15. Other:	0	1	2	3	4	1	2	3

STEREOTYPED BEHAVIOR

Generic definition: *Stereotyped behaviors look unusual, strange, or inappropriate to the average person. They are voluntary acts that occur repeatedly in the same way over and over again, and they are characteristic for that person. However, they do NOT cause physical damage.*

	Never	Frequency				Degree of Problem		
		monthly	weekly	daily	hourly	slight	moderate	severe
16. Rocking back and forth	0	1	2	3	4	1	2	3
17. Sniffing objects	0	1	2	3	4	1	2	3
18. Spinning own body	0	1	2	3	4	1	2	3
19. Waving or shaking arms	0	1	2	3	4	1	2	3
20. Rolling head	0	1	2	3	4	1	2	3

21. Whirling, turning around on spot	0	1	2	3	4	1	2	3
22. Engaging in repetitive body movements	0	1	2	3	4	1	2	3
23. Pacing	0	1	2	3	4	1	2	3
24. Twirling things	0	1	2	3	4	1	2	3
25. Having repetitive hand movements	0	1	2	3	4	1	2	3
26. Yelling and screaming	0	1	2	3	4	1	2	3
27. Sniffing own body	0	1	2	3	4	1	2	3
28. Bouncing around	0	1	2	3	4	1	2	3
29. Spinning objects	0	1	2	3	4	1	2	3
30. Having bursts of running around	0	1	2	3	4	1	2	3
31. Engaging in complex hand and finger movements	0	1	2	3	4	1	2	3
32. Manipulating objects repeatedly	0	1	2	3	4	1	2	3
33. Exhibiting sustained finger movements	0	1	2	3	4	1	2	3
34. Rubbing self	0	1	2	3	4	1	2	3
35. Gazing at hands or objects	0	1	2	3	4	1	2	3
36. Maintaining bizarre body postures	0	1	2	3	4	1	2	3
37. Clapping hands	0	1	2	3	4	1	2	3
38. Grimacing	0	1	2	3	4	1	2	3
39. Waving hands	0	1	2	3	4	1	2	3
40. Other	0	1	2	3	4	1	2	3

AGGRESSIVE/DESTRUCTIVE BEHAVIOR

Generic definition: *Aggressive or destructive behaviors are offensive actions or deliberate overt attacks directed towards other individuals or objects. They occur repeatedly in the same way over and over again, and they are characteristic for that person.*

	Never	Frequency			Degree of Problem			
		monthly	weekly	daily	hourly	slight	moderate	severe
41. Hitting others	0	1	2	3	4	1	2	3
42. Kicking others	0	1	2	3	4	1	2	3
43. Pushing others	0	1	2	3	4	1	2	3
44. Biting others	0	1	2	3	4	1	2	3
45. Grabbing and pulling others	0	1	2	3	4	1	2	3

46. Scratching others	0	1	2	3	4	1	2	3
47. Pinching others	0	1	2	3	4	1	2	3
48. Spitting on others	0	1	2	3	4	1	2	3
49. Being verbally abusive with others	0	1	2	3	4	1	2	3
50. Destroying things (e.g., rips clothes, throws chairs, smashes tables)	0	1	2	3	4	1	2	3
51. Being mean or cruel (e.g., grabbing toys or food from others, bullying others)	0	1	2	3	4	1	2	3
52. Other:	0	1	2	3	4	1	2	3

Appendix 3

The Reiss Scales for Children's Dual Diagnosis

This test presents a list of maladaptive behaviours that could create problems in the lives of children with special needs. Each item on the list is defined. A few examples are given to help you understand the meaning of the definition. Your task is to read each item and tell us if you think that the item is currently **NO PROBLEM**, a **PROBLEM**, or a **MAJOR PROBLEM** in the child's life. Please keep in mind that we do not want to know simply if the behaviour occurs; what we would like is your opinion if the problem occurs with sufficient frequency, with sufficient intensity, or under sufficiently strange or inappropriate circumstances, so that the behaviour category is a problem or a major problem in the child's life.

Rating Scale

NO PROBLEM. Use this rating if any of the following are true:

1. The behaviour category does not apply to the child you are rating. For example, the category of "lying" does not apply to a child who is non-verbal.
2. The child you are evaluating does not engage in the behaviour.
3. The behaviour does not occur with sufficient frequency, intensity, or severity to be considered a current problem in the life of the child you are evaluating.

PROBLEM. Use this rating if one or more of the following are true:

1. The behaviour causes a significant degree of discomfort and/or suffering for the child being evaluated.
2. The behaviour interferes with the child's social functioning.
3. The behaviour interferes with the child's school functioning.
4. The behaviour occurs often or with unusual degree of severity.

MAJOR PROBLEM. Use this rating if one or more of the following are true:

1. The behaviour causes a great deal of discomfort and/or suffering for the child you are evaluating.
2. The behaviour occurs with very high frequency or intensity.
3. The behaviour significantly interferes with the child's social adjustment.
4. The behaviour causes placement in a restrictive environment or increases the need for supervision.

1. Afraid of strangers. Becomes fearful in the presence of adult strangers. e.g. resists going near an unfamiliar adult even when encouraged to do so under appropriate circumstances, cries when meeting an adult for the first time, cries in a crowd. No Problem Problem Major Problem

2. Angry. Frequently feels hostile or mad. Example: gets mad easily, argues a lot, interrupts others when ignored. No Problem Problem Major Problem

3. Anxious. Appears nervous or tense. e.g. nervous, overreacts to unexpected sounds or events, vigilant, worried. No Problem Problem Major Problem

4. Avoids by Illness. False sickness, disability, or pain in order No Problem Problem Major Problem

- to avoid something he/she does not want to do. e.g. says he/she has a stomach-ache in order to avoid going to school, says he/she has a headache in order to avoid cleaning up room.
- 5. Avoids Peers.** Dislikes interacting with other children. e.g. prefers to play alone, avoids groups, parallel play only, pushes/hits others when approached. No Problem Problem Major Problem
- 6. Bizarre Ideas.** Expressed strange ideas. e.g. says that he/she is a sailor, says that he/she should collect as many rocks as possible. No Problem Problem Major Problem
- 7. Blank Stares.** Appears expressionless and emotionless. e.g. sometimes appears to be in a trance, gazes off into space. No Problem Problem Major Problem
- 8. Bodily Complaints.** Complains about aches and pains. e.g. headaches, stomach-aches, dizziness, constipation, diarrhoea, unexplained recurrent pains. No Problem Problem Major Problem
- 9. Bonding Problem.** Child or infant has not formed normal emotional attachments with parents/caregivers. e.g. does not seek closeness if caretakers enters room, does not calm when held by parents, does not respond to affection from parents/caretakers. No Problem Problem Major Problem
- 10. Bullies Others.** Controls others with threats, verbal abuse, or actual physical attack. e.g. intimidates smaller or weaker children, bosses around smaller or weaker children. No Problem Problem Major Problem
- 11. Changes In Sleep Behaviour.** A change in usual sleep habits. e.g. recent trouble falling asleep, wakes up in the middle of the night, has trouble waking in mornings. No Problem Problem Major Problem
- 12. Communication Problem.** Marked difficulty in communicating with others. E.g. makes up and uses own words for things, no mode of communication, abnormal gestures, marked speech problem, echolalia, stuttering. No Problem Problem Major Problem
- 13. Confusing Speech.** Poorly related or bizarre ideas or thought. e.g. speech makes no sense, thinking is hard to follow, expresses strange ideas, thought jump from one topic to another. No Problem Problem Major Problem
- 14. Crying Spells.** Periodic bouts of sobbing. E.g. easily moved to tears, cries more often than most children, cries for no apparent reason. No Problem Problem Major Problem
- 15. Destructive.** Deliberately damages property. E.g. breaks windows, deliberately destroys furniture, throws objects, turns over furniture. No Problem Problem Major Problem

- 16. Disobedient.** Does not follow rules or directions given by people in authority. E.g. does not listen to teacher, does not follow rules of group home/residence, does not follow simple requests. No Problem Problem Major Problem
- 17. Distracted.** Attention to a task is easily interrupted by extraneous or irrelevant stimuli. Example: short attention span, has trouble concentrating. No Problem Problem Major Problem
- 18. Enuresis/Encopresis.** A child beyond the age of toilet training with inadequate bladder or bowel control. e.g. bed wetting, urinating on the floor, defecating in pyjamas or pants. No Problem Problem Major Problem
- 19. Excessive Need For Reassurance.** Frequently needs to be told that things are okay. E.g. excessive need to be told that he/she is loved or liked, excessive need to be told that he/she is doing a good job, repeatedly needs to be told that time of a schedule event or reassured that it will occur. No Problem Problem Major Problem
- 20. Excessive Sensitivity To Criticism.** Excessive or inappropriate reactions to criticism. E.g. reacts to failure by crying, quits easily, become angry, becomes angry No Problem Problem Major Problem
- 21. Fearful.** Afraid of many objects or situations. E.g. afraid to go places, afraid to try new activities, afraid of many different things. No Problem Problem Major Problem
- 22. Feels Unloved.** Has perceptions that parents or significant others do not love or care about him/her. E.g. says that parents/caretakers do not love him/her, says that nobody cares about him/her, says that parents/caretakers love others (e.g. brother or sisters) more. No Problem Problem Major Problem
- 23. Gaze Avoidance.** Actively avoids eye contact. E.g. infrequent eye contact with others, becomes upset when face-to face contact is forced. No Problem Problem Major Problem
- 24. Hallucinations.** Experiences things that are not there. E.g. hears voices. Hears sounds, has visions, feels strange bodily sensations. No Problem Problem Major Problem
- 25. Headaches.** Complains about aches and pains in the head. E.g. says head hurts, has migraine headaches, has tension headaches. No Problem Problem Major Problem
- 26. Impatient.** Needs/demands must be met immediately. E.g. demanding, can't wait his/her turn, easily frustrated. No Problem Problem Major Problem
- 27. Impulsive.** Reacts quickly without first thinking about the likely consequences. E.g. makes decisions quickly, quick-tempered. No Problem Problem Major Problem
- 28. Inattentive.** Pays little attention to people or to events No Problem Problem Major Problem

around him/her. E.g. pays little attention when spoken to, seems "spaced out".

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| 29. Involuntary Motor movements. Repetitive movements beyond the control of the person. E.g. excessive blinking, strange motor movements, frequent shrugs, handflapping. | No Problem Problem Major Problem |
| 30. Irritable. Easily annoyed or provoked. E.g. easily frustrated, becomes angry over minor annoyances, easily offended, feelings are hurt easily. | No Problem Problem Major Problem |
| 31. Isolated. Spends a lot of time alone. E.g. has no friends, plays alone, is ignored or avoided by other children. | No Problem Problem Major Problem |
| 32. Lacks Enjoyment. Does not seem to enjoy things anymore. E.g. has no fun, does not want to play anymore, does not want to do much of anything. | No Problem Problem Major Problem |
| 33. Lies. Habitually says things that he/she knows are false or misleading. E.g. lies about getting into fights, fabricates incredible tales, lies about being late. | No Problem Problem Major Problem |
| 34. Negative Self-Image. Dislikes self. E.g. he/she is stupid, says he/she is a bad person, says he/she is ugly. | No Problem Problem Major Problem |
| 35. Obese. Excessively overweight. Example: perceived by others as being fat, eats too much. | No Problem Problem Major Problem |
| 36. Object Attachment. Strong and persistent attachments to a particular object. E.g. Often wants to hold a particular ball, searches for missing objects, likes to carry a key chain and gets upset when the key chain cannot be found. | No Problem Problem Major Problem |
| 37. Overactive. Excessive movement to the point where the person has difficulty staying still. E.g. appears to be in constant motion, excessive physical movement, pacing, constantly changing activity. | No Problem Problem Major Problem |
| 38. Pessimistic. Has a negative view of the future. E.g. negative outlook, lacks hope, expects the worst, negative thinking. | No Problem Problem Major Problem |
| 39. Physically Aggressive. Physically attacks others. E.g. fights, spits on others, hits others. | No Problem Problem Major Problem |
| 40. Pica. Tendency to eat non-edible objects. E.g. eats dirt, eats paint chips, eats paper, drinks cleaner solution. | No Problem Problem Major Problem |
| 41. Rebellious. Defies authority and/or resists control from adults. e.g. defiant, refuses to co-operate with adults, hostile toward authority figures. | No Problem Problem Major Problem |

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| 42. Runs Away. Leaves without permission and without informing other people. E.g. runs away from home, residential facility, runs away from school. | No Problem Problem Major Problem |
| 43. Sad. Displays frequent or excessive feelings of unhappiness. e.g. often gives appearance of unhappy child, has bouts of crying, rarely smiles. | No Problem Problem Major Problem |
| 44. Seeks Medical Care. Frequently asks for or seeks out medical attention. E.g. asks for medicine, often needs medical care for one thing after another. | No Problem Problem Major Problem |
| 45. Self-Injury. Repeatedly injures body on purpose. E.g. bites arm, hits self repeatedly, bangs head repeatedly. | No Problem Problem Major Problem |
| 46. Self-Stimulatory Behaviour. Repetitive movements that are performed frequently and appear to be non-functional. E.g. body-rocking, object twirling, head rocking. | No Problem Problem Major Problem |
| 47. Separation Anxiety. Afraid of being away from parent/caretaker. e.g. body-rocking, object-twirling, head-rocking. | No Problem Problem Major Problem |
| 48. Sets Fire. Deliberately starts fires. Example: sets fire to room, sets fire to schools. | No Problem Problem Major Problem |
| 49. Sexual Problem. Repeatedly performs sexual behaviours that are socially disapproved. e.g. sexual expression at in appropriate times or places, masturbates in public. | No Problem Problem Major Problem |
| 50. Shy. Uncomfortable in the presence of other people. e.g. dislikes being the centre of attention, bashful, ill at ease in groups, dislikes meeting new people. | No Problem Problem Major Problem |
| 51. Social Inadequacies. Has difficulty relating to peers in appropriate or satisfying ways. e.g. has no friends, tends to be disliked, insensitive to the feelings of other people. | No Problem Problem Major Problem |
| 52. Steals. Takes property that belongs to others. e.g. takes classmate's possessions, takes money for others. | No Problem Problem Major Problem |
| 53. Stomach aches. Complains about stomachaches. e.g. says stomach is up[set, feels nauseous, complains of gassy stomach. | No Problem Problem Major Problem |
| 54. Strange Behaviour. Engages in behaviour that impresses many observers as unusual, peculiar, strange, or bizarre. e.g. hoards food in pockets or under en, unusually wears several layers of clothes regardless of weather, always mutters things to self. | No Problem Problem Major Problem |

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| 55. Suicidal Statements. Thinks about, attempts, or threatens to kill himself/herself. e.g. says that he/she would like to die, intentionally cuts or hurts self, tries to get run over by cars. | No Problem Problem Major Problem |
| 56. Temper Tantrums. Angry outbursts when frustrated or disappointed. e.g. shouts and yells when not given in to, has outburst when asks to do something he/she does not want to do. | No Problem Problem Major Problem |
| 57. Uncompleted Activities. Marked tendency not to finish things. e.g. usually does not finish, goes from one uncompleted activity to another. | No Problem Problem Major Problem |
| 58. Unusual Vocalizations. Makes strange or unusual sounds. e.g. grunts, barking noises, whispers words, sudden anger or swear words when not obviously angry. | No Problem Problem Major Problem |
| 59. Verbally Abusive. Threatens or insults other people e.g. taunts, insults, threatens others, makes fun of other people, yells or shouts at others. | No Problem Problem Major Problem |
| 60. Withdrawn. Avoids personal contact with other people. e.g. excessively shy, doesn't participate in group activities, prefers to be alone, socially isolated. | No Problem Problem Major |

Appendix 4

Nisonger Child Behaviour Rating Form -social competence sub scales

Please describe your child's behaviour as it was at home over the last month.

1. Accepted redirection	0	1	2	3
2. Expressed ideas clearly	0	1	2	3
3. Followed rules	0	1	2	3
4. Initiated positive interactions	0	1	2	3
5. Participated in group activities	0	1	2	3
6. Resisted provocation, was tolerant	0	1	2	3
7. Shared with or helped others	0	1	2	3
8. Stayed on task	0	1	2	3
9. Was cheerful or happy	0	1	2	3
10. Was patient, able to delay	0	1	2	3

Appendix 5

The Parent and Family subscale of the Questionnaire on Resources and Stress

The following statements deal with your feelings about your child with Angelman Syndrome. There are many blank spaces on the questionnaire (_____). Imagine the name of your child with Angelman Syndrome in each of these blank spaces. Please give your honest feelings and opinions. Respond to all of the statements, even if they do not seem to apply. If it is difficult to decide "true" or "false", answer in terms of what you or your family do *most* of the time.

1. Other members of the family have to do without things because of _____.	True	False
2. Our family agrees on important matters.	True	False
3. The constant demands for care for _____ limit growth and development of someone else in the family.	True	False
4. I have given up things I really wanted to do in order to care for _____.	True	False
5. _____ is able to fit into the family group.	True	False
6. In the future, our family's social life will suffer because of the increased responsibilities and financial stress.	True	False
7 I can go to visit friends whenever I want.	True	False
8. Taking _____ on holiday spoils the pleasure for the whole family.	True	False
9. The family does as many things together now as we ever did.	True	False
10. There are many places where we can enjoy ourselves as a family when _____ comes along	True	False
11. There is a lot of anger and resentment in our family	True	False
12. The constant demands to care for _____ limit my growth and development	True	False
13. I feel sad when I think of _____.	True	False
14. Caring for _____ puts a strain on me.	True	False
15. Members of our family get to do the same kinds of things other families do.	True	False

Appendix 6

The Positive Contributions Scale from the Kansas Inventory of Parental Perceptions (KIPP)

MY CHILD _____ IS:

The blank space after the word “child” is there to remind you to think only of your child with special needs when you answer each statement. Read each statement and circle the one response that best describes how much you agree or disagree with each statement.

Part A

MY CHILD _____ IS:

	Strongly Disagree	Disagree	Agree	Strongly Agree
1. the reason I attend religious services more frequently.	1	2	3	4
2. why I met some of my best friends.	1	2	3	4
3. the reason my life has better structure.	1	2	3	4
4. why I am a more responsible person.	1	2	3	4
5. the reason I've learned to control my temper.	1	2	3	4
6. responsible for my learning patience.	1	2	3	4
7. responsible for my increased awareness of people with special needs.	1	2	3	4
8. fun to be around.	1	2	3	4
9. the reason I am more realistic about my job.	1	2	3	4
10. responsible for my being more aware and concerned for the future of mankind.	1	2	3	4
11. kind and loving.	1	2	3	4
12. helpful to other family members, which saves time and energy for me.	1	2	3	4
13. a source of pride because of his/her artistic accomplishments.	1	2	3	4

Part B

I CONSIDER MY CHILD _____ TO BE:

14. what gives me common ground with other parents.	1	2	3	4
15. helpful without having to be asked.	1	2	3	4
16. responsible for my increased sensitivity to people.	1	2	3	4
17. what gives our family a sense of continuity – a sense of history.	1	2	3	4
18. the reason I am more productive.	1	2	3	4
19. an advantage to my career.	1	2	3	4
20. the reason I budget my time better.	1	2	3	4
21. the reason I am able to cope better with stress and problems.	1	2	3	4
22. very affectionate.	1	2	3	4
23. what makes me realise the importance of planning for my family's future.	1	2	3	4
24. able to use good judgement.	1	2	3	4

25. a great help around the house. 1 2 3 4

Part C

THE PRESENCE OF MY CHILD _____:

- 26. is an inspiration to improve my job skills. 1 2 3 4
- 27. helps me understand people who are different. 1 2 3 4
- 28. is a source of pride because of his/her athletic achievements. 1 2 3 4
- 29. cheers me up. 1 2 3 4
- 30. confirms my faith in God. 1 2 3 4
- 31. gives a new perspective to my job. 1 2 3 4
- 32. renews my interest in participating in different activities. 1 2 3 4
- 33. is very uplifting. 1 2 3 4
- 34. is a reminder that all children, including those with special needs, need to be loved. 1 2 3 4
- 35. is a reminder that everyone has a purpose in life. 1 2 3 4
- 36. makes us more in charge of ourselves as a family. 1 2 3 4
- 37. helps me take things as they come. 1 2 3 4

Part D

BECAUSE OF MY CHILD _____:

- 38. my circle of friends has grown larger. 1 2 3 4
- 39. I have someone who shares responsibility for doing several tasks around the house. 1 2 3 4
- 40. my social life has expanded by bringing me into contact with other parents. 1 2 3 4
- 41. I am more compassionate. 1 2 3 4
- 42. I learned about mental retardation. 1 2 3 4
- 43. my family is more understanding about special problems. 1 2 3 4
- 44. I am grateful for each day. 1 2 3 4
- 45. our family has become closer. 1 2 3 4
- 46. I am more sensitive to family issues. 1 2 3 4
- 47. I have learned to adjust to things I cannot change. 1 2 3 4
- 48. my other children have learned to be aware of people's needs and their feelings. 1 2 3 4
- 49. I have many unexpected pleasures. 1 2 3 4
- 50. I am more accepting of things. 1 2 3 4

Appendix 7

Hospital Anxiety and Depression Scale

This questionnaire focuses on how you feel about things. Please read each item and circle the reply underneath the item which comes closest to how you have been feeling in the past week. Do not take too long over your replies; your immediate reaction to each item will probably be more accurate than a long thought-out response.

1. I feel tense or "wound up"

Most of the time	A lot of the time	Occasionally, from time to time	Not at all
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2. I still enjoy the things I used to enjoy

Definitely as much	Not quite so much	Only a little	Hardly at all
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3. I get a sort of frightened feeling as if something awful is about to happen

Very definitely and quite badly	Yes, but not too badly	A little, but it doesn't worry me	Not at all
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4. I can laugh and see the funny side of things

As much as I always could	Not quite so much now	Definitely not so much now	Not at all.
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5. Worrying thoughts go through my mind

A great deal of the time	A lot of the time	From time to time but not too often	Only occasionally
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6. I feel cheerful

Not at all	Not often	Sometimes	Most of the time
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7. I can sit at ease and feel relaxed

Definitely	Usually	Not often	Not at all
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8. I feel as if I am slowed down

Nearly all the time	Very often	Sometimes	Not at all
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9. I get a sort of frightened feeling like "butterflies" in the stomach

Not at all	Occasionally	Quite often	Very often
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10. I have lost interest in my appearance

Definitely	I don't take as much care as I should	I may not take quite as much care	I take just as much care as ever
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11. I feel restless as if I have to be on the move

Very much indeed	Quite a lot	Not very much	Not at all
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12. I look forward with enjoyment to things

As much as I ever did	Rather less than I used to	Definitely less than I used to	Hardly at all
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13. I get sudden feelings of panic

Very often indeed	Quite often	Not very often	Not at all
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14. I can enjoy a good book, radio or TV programme

Often	Sometimes	Not often	Very seldom
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Appendix 8

Positive Affect Scale (PAS)

This scale consists of a number of words that describe different feelings and emotions. Read each item and then circle one of the responses. **Indicate to what extent you have felt this in the past week.**

	Very slight or not at all	A little	Moderate	Quite a bit	Extremely
1. Interested	1	2	3	4	5
2. Excited.	1	2	3	4	5
3. Strong.	1	2	3	4	5
4. Enthusiastic	1	2	3	4	5
5. Proud	1	2	3	4	5
6. Alert	1	2	3	4	5
7. Inspired.	1	2	3	4	5
8. Determined	1	2	3	4	5
9. Attentive	1	2	3	4	5
10. Active	1	2	3	4	5

Appendix 9

Satisfaction with Life Scale

Below are five statements that you may agree or disagree with. Read each one and circle the response that best describes how strongly you agree or disagree.

	Strongly Disagree	Disagree	Slightly Disagree	Neither Agree or Disagree	Slightly Agree	Agree	Strongly Agree
1. In most ways, my life is close to my ideal.	1	2	3	4	5	6	7
2. The conditions of my life are excellent.	1	2	3	4	5	6	7
3. I am completely satisfied with my life.	1	2	3	4	5	6	7
4. So far I have got the most important things I want in life.	1	2	3	4	5	6	7
5. If I could live my life over again, I would change nothing.	1	2	3	4	5	6	7

Appendix 10

The Golombok Rust Inventory of Marital State (GRIMS)

You and Your Partner

Please read each statement and decide which response best describes how you feel about your partner. Please do not discuss any of the responses with your partner.

	Strongly Disagree	Disagree	Agree	Strongly Agree
1. My partner is usually sensitive to and aware of my needs	1	2	3	4
2. I really appreciate my partner's sense of humour	1	2	3	4
3. My partner doesn't seem to listen to me any more	1	2	3	4
4. My partner has never been disloyal to me	1	2	3	4
5. I would be willing to give up my friends if it meant saving our relationship	1	2	3	4
6. I am dissatisfied with our relationship	1	2	3	4
7. I wish my partner was not so lazy and didn't keep putting things off	1	2	3	4
8. I sometimes feel lonely even when I am with my partner	1	2	3	4
9. I am with my partner if my partner left me life would not be worth living	1	2	3	4
10. We can "agree to disagree" with each other	1	2	3	4
	1	2	3	4
11. It is useless carrying on with a marriage beyond a certain point				
12. We both seem to like the same things	1	2	3	4
13. I find it difficult to show my partner that I am feeling affectionate	1	2	3	4
14. I never have second thoughts about our relationship	1	2	3	4
15. I enjoy just sitting and talking with my partner	1	2	3	4
16. I find the idea of spending the rest of my life with my partner rather boring	1	2	3	4
17. There is always plenty of "give and take" in our relationship	1	2	3	4
18. We become competitive when we have to make decisions	1	2	3	4

19. I no longer feel I can really trust my partner	1	2	3	4
20. Our relationship is still full of joy and excitement	1	2	3	4
21. One of us is continually talking and the other is usually silent	1	2	3	4
22. Our relationship is continuously evolving	1	2	3	4
23. Marriage is really more about security and money than about love	1	2	3	4
24. I wish there was more warmth and affection between us	1	2	3	4
25. I am totally committed to my relationship with my partner	1	2	3	4
26. Our relationship is sometimes strained because my partner is always correcting me	1	2	3	4
27. I suspect we may be on the brink of separation	1	2	3	4
28. We can always make up quickly after an argument	1	2	3	4

Appendix 11

The Family Satisfaction Scale

Your View of Your Family

This questionnaire is concerned with family satisfaction. Please circle how satisfied you are with the following items.

How satisfied are you:

	Dissatisfied	Somewhat Satisfied	Generally Satisfied	Very Satisfied	Extremely Satisfied
1. with how close you feel to the rest of your family?	1	2	3	4	5
2. with your ability to say what you want in your family?	1	2	3	4	5
3. with your family's ability to try new things?	1	2	3	4	5
4. with how fair the criticism is in your family?	1	2	3	4	5
5. with the amount of time you spend with your family?	1	2	3	4	5
6. with the way you talk together to solve family problems?	1	2	3	4	5
7. with your freedom to be alone when you want to?	1	2	3	4	5
8. with how strictly you stay with who does what chores in your family?	1	2	3	4	5
9. with your family's acceptance of your friends?	1	2	3	4	5
10. with how clear is it what your family expects of you?	1	2	3	4	5
11. with how often you make decisions as a family, rather than individually?	1	2	3	4	5
12. with the number of fun things your family does together?	1	2	3	4	5

Positive Gain Scales

The following questions ask you about your feelings associated with raising a child with Angelman Syndrome. Please mark the answer that comes closest to describing how you feel. Your first reaction to each question should be your answer.

	Strongly Agree	Agree	Not sure	Disagree	Strongly Disagree
1. Since having this child I feel I have grown as a person.	SA	A	NS	D	SD
2. Having this child has helped me to learn new things/skills.	SA	A	NS	D	SD
3. Raising this child helps putting life into perspective.	SA	A	NS	D	SD
4. Since having this child, my family has become closer to one another.	SA	A	NS	D	SD
5. Since having this child my family has become more tolerant and accepting	SA	A	NS	D	SD
6. Since having this child I have become more determined to face up to challenges.	SA	A	NS	D	SD
7. Since having this child I have a greater understanding of other people.	SA	A	NS	D	SD

Genetic Syndromes Stressors Scale (GSSS)

The following questions are about specific sources of stress relating to raising a child with a rare genetic syndrome. Read each item and then circle the response that best describes your experiences. If the item does not relate to your experiences, please circle '0'.

How stressful have you found the following issues in the past 6 months?

	Not at all stressful	A little stressful	Moderately stressful	Extremely stressful
1. Not having access to professionals who have knowledge about my child's condition	0	1	2	3
2. People staring when I go out in public with my Child	0	1	2	3
3. Getting my child's complex needs met through social services	0	1	2	3
4. The large amount of effort required to help my child reach developmental milestones (e.g. sitting up, self-feeding)	0	1	2	3
5. Having to be constantly vigilant about my child's state of health in case of a sudden change	0	1	2	3
6. Going to see professionals who are not knowledgeable about my child's genetic syndrome	0	1	2	3
7. Arranging care (e.g. babysitting, respite) that is suitable for my child	0	1	2	3
8. An educational placement that does not meet all of my child's needs	0	1	2	3
9. Sleep deprivation, due to my child's sleeping patterns	0	1	2	3
10. A genetic diagnosis causing tension within the immediate and extended family	0	1	2	3
11. Not being able to fully relax at home, as I need to attend to my child 24 hours a day	0	1	2	3
12. Having to explain my child's condition to new people I meet	0	1	2	3
13. Having to make extensive preparations for my child before leaving the house	0	1	2	3
14. Worrying about the future for my child because of the lack of specialist services once they reach adulthood	0	1	2	3

Initial contact letter and Information sheets

14a: Three syndromes project initial contact letter to parents

14b: Initial information sheet to parents

Appendix 14a: Three syndromes project initial contact letter to parents

The Three Syndromes Study
The Angelman, Cri du Chat and Cornelia de Lange Syndrome Support Groups
Funded by The Big Lottery to tackle behaviour problems and support families



cri du chat



Dear X,

We are writing to inform you of a new research project that is being conducted at the University of Birmingham in collaboration with the University of Wales, Bangor and the Institute of Psychiatry, King College London. The research project is being conducted in association with the Cornelia de Lange Syndrome Foundation (UK & Ireland), Cri du Chat Syndrome Support Group and the Angelman Syndrome Support, Research and Education trust and is supported by the Big Lottery Fund.

The research aims to improve our understanding of the role of factors within the environment that may impact on self-injurious and aggressive behaviours commonly associated with Cornelia de Lange, Cri du Chat and Angelman syndromes. The study will also consider factors that are related to family well-being and adjustment. This is an important area of study but has rarely been attended to within the literature.

We have selected seventy-five individuals from our database of families who we feel may be well suited to participate in this research project. We would like to make telephone contact with you in the next 7 days to discuss further the possibility of you and your son/daughter X taking part in this study.

There is an information sheet enclosed that gives you some more detailed information 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 that you care for.

Please take the time to read the enclosed information sheet. If you are unclear about any aspect of the study or have any queries then please contact Professor Chris Oliver by telephone: 0121 414 4909, email: c.oliver@bham.ac.uk or at the above address.

Thank you for your time and continued support for our research at the University of Birmingham. We look forward to speaking to you in the next 7 days. .

Yours sincerely,

Professor Chris Oliver (Project Director)

The Three Syndromes Study
 The Angelman, Cri du Chat and Cornelia de Lange Syndrome Support Groups
 Funded by The Big Lottery to tackle behaviour problems and support families



cri du chat



Introduction to the research and invitation to take part:

We have selected you and your child/person you care for as potential participants in a new study being conducted at the University of Birmingham, in collaboration with the University of Wales, Bangor and the Institute of Psychiatry, Kings College London.

The research project is being conducted in association with the Cornelia de Lange Syndrome Foundation (UK & Ireland), Cri du Chat Syndrome Support group and the Angelman Syndrome Support, Research and Education Trust and is supported by the Big Lottery Fund.

The study aims to improve our understanding of the role of factors within the environment that may impact on self injurious and aggressive behaviours commonly associated with Cornelia de Lange, Cri du Chat and Angelman syndromes. We will also examine the factors that are related to family well-being within these syndrome groups. We hope that greater understanding of the behaviours associated with these syndrome groups will help to support social inclusion, develop better intervention and management strategies for families and improve the health and well-being of affected individuals and their families.

What does it involve?

Participation in the research project will involve the following:

- You will be asked to complete 2 brief questionnaire packs in order to provide us with some background information about your child/person you care for and their behaviour.
- We would like to take some time to talk to you about your child's/person you care for's behaviour. Some of this will be done over the phone but we will also visit you at home.
- We will visit your child/person you care for at their school or day centre 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. These different social situations and activities will be presented to your child/person you care for by two members of the research team.
- We will ask your child's/person you care for's teacher/key worker to complete some brief questionnaires and will ask them to take some time to talk to us about your child's/person you care for's behaviour at school.

An example of the timetable for collecting the above information from you, your child/person you care for and their teacher is shown below.

Stage One:	Return consent form
Stage Two:	Complete questionnaire pack.
Stage Three:	Complete phone interview
Stage Four:	Two research workers will visit you at home to talk about your

	child's/person you care for's behaviour.
Stage Five:	Two research workers will visit your child/person you care for at school for the day.
Stage Six:	You will receive a detailed individual feedback form about your child's/person you care for's assessments.

How will behaviours be observed and recorded?

- When we visit your child/person you care for at school/day centre, we will carry out short observations with them in different social situations and during a series of games and activities. Video recordings of your child/person you care for during these situations and activities will be made and stored. This allows us to return to the video recordings for detailed analysis of information and means that we can check the accuracy of our observations.
- When videotapes are not used they will be stored in a locked filing cabinet.
- In the future we may contact you again to ask if you would be happy for us to use video recordings for teaching purposes. However, agreeing to participate in this study does not mean that you will be obliged to give your permission for the use of these video recordings in the future.
- The privacy and dignity of your child/person you care for will be respected at all times and video recordings will not take place if there is evidence that the observations are causing distress.
- You may ask to see a copy of the video recordings of your child/person you care for.
- Video recordings may only be viewed by legal guardians or individuals providing a service to the person you care for and members of the research team working on this project.
- Information identifying your child/ person you care for will not be stored on or with the tape.
- The University of Birmingham will hold the copyright for the video recordings in order that the confidentiality of the recordings of your child/person you care for be protected. However, this does not mean that the University of Birmingham will have the right to edit, copy or use the videos for teaching purposes without your written permission.
- We may contact you again in the future to ask your permission to use the video recordings for teaching purposes. At that stage you will be able to decide whether or not you are happy for the videos to be used for these purposes. However, agreeing to participate in this study does not mean that you will be obliged to give your permission for the use of these video recordings in the future.

Consent:

After our phone call, if you decide to become involved in the project then you will be required to complete a consent form and return this us. If your child/person you care for is over 16 and is able to give consent themselves they may complete the form.

Withdrawal:

Should you or the person you care for decide that you no longer wish to be involved in the research; you are free to withdraw your participation at anytime during the study and for a period of three months after the data collection with yourselves has been completed. If you decide to do so, information that you have provided in this time can also be

withdrawn and destroyed without you giving reason. This will not restrict access to other services and will not affect the right to treatment.

Confidentiality:

All information collected will be kept on a confidential database that is only accessible to those working on the project. In the unlikely event of any evidence of abuse being identified, this information will be disclosed by the research workers. All personal details will be kept separately from the information collected and your child/person you care for will be identifiable by a code throughout the study to ensure anonymity. 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/the person they care for.

A summary of the project's findings will be circulated to anyone involved who wishes to see a copy. Any requests for advice concerning your child/person you care for will be referred to Professor Chris Oliver, Clinical Psychologist. It is possible that you may be invited to participate in further research after the study. However, consenting to participate in this study does not mean that you are obliged to do so.

Who has reviewed the study?

This research project has been reviewed and approved by The University of Birmingham, School of Psychology Research Ethics Committee (0121 424 0594), the University of Bangor Ethics Committee (01248 382211 ext 8771) and the Kings College London Ethics Committee (0207 848 4020; ref: CREC/06/07-190)

Any concerns of queries?

If you are unclear about any aspect of the study or have any questions, please do not hesitate to contact Professor Chris Oliver by telephone: 0121 414 4909, by email: c.oliver@bham.ac.uk or at the following address:

Professor Chris Oliver
School of Psychology
University of Birmingham
Edgbaston
Birmingham, B15 2TT

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

Supported by
 **The National Lottery***
through the Big Lottery Fund



There are just a few questions we need to ask you to make sure that X is suitable for this research project:

1. Does X currently show any self-injurious behaviours such as head banging or biting?
 - a. If so, what does this behaviour look like? (get them to describe the basic topography such as head banging on the floor)
 - b. How frequently would you say this behaviour occurs typically? (weekly, daily, more frequently, less frequently? If it is unclear, prompt with the question- 'if nothing changed and we watched X, when would we definitely see the behaviour?' By this time tomorrow, by next week, in the next few hours etc)
 - c. Does the behaviour currently cause tissue damage? (ie bruising, bleeding, loss of hair etc)
 - d. What response is typically required? (ie say no, block hand, splints etc)

2. Does X currently show any physically aggressive behaviour? (*NB: parents may not define behaviour as aggressive in function, ie the child might not intentionally MEAN to be aggressive, we are interested in the topography rather than function at this stage- does it happen, regardless of why*).
 - a. If so, what form does this usually take? (kicking, punching, hair pulling etc)
 - b. How frequently would you say this behaviour occurs typically? (weekly, daily, more frequently, less frequently? If it is unclear, prompt with the question- if nothing changed and we watched X, when would we definitely see the behaviour? By this time tomorrow, by next week, in the next few hours etc)
 - c. Who is the behaviour usually directed towards? (ie parents, siblings, anyone etc)
 - d. Does this behaviour cause injury to others or severe disruption to X's daily life? (ie effect on day placement, trips into community etc)

OK= parent indicates that self-injurious or aggressive behaviour is occurring at least daily.

NO= parent indicates that self-injurious or aggressive behaviour is not occurring or is occurring less than once per day.

Recruitment flyer “Three syndromes” project



The 3 Syndromes Study Research Team is looking for you!

The University of Birmingham research team has started a new research project about challenging behaviour and family wellbeing in children and adults with Cornelia de Lange, Angelman and Cri du Chat syndromes. Our research team has now expanded to include research teams in both London and North Wales so that we are now able to get in touch with more families and children around the UK.

In this project we are inviting children and adults aged between *2 and 15* years who are showing aggressive or self-injurious behaviour *at least once a day* to take part in the study. At this stage we would simply like to inform you of the study and ask any families of children and adults who are engaging in these behaviours to get in touch with us.

- **How do we define aggressive behaviour?** Aggressive behaviour includes any behaviour which may cause physical discomfort or possible harm to another person including hair pulling, grabbing, scratching, pushing, kicking, biting, hitting etc. It may well be that the person doesn't mean to hurt others or is too small to hurt others. For this project we would still be interested in hearing from you.
- **How do we define self-injurious behaviour?** Self-injurious behaviour includes any behaviour which may cause physical discomfort (including reddening of the skin or bruising) or harm to the person such as picking, biting, tapping, hitting, banging, scratching etc.

If the person you care for shows either or both of the above behaviours at least once a day *or* they show something like self-injury or aggression but you're not sure if they could be included, then please contact Professor Chris Oliver at the University of Birmingham on: 0121 414 4908, via email: c.oliver@bham.ac.uk or post: Professor Chris Oliver, School of Psychology, University of Birmingham, Edgbaston, B15 2TT. *Responding to this leaflet does not in any way commit you to participation in the study. We will give you some more information, you can discuss the project with us and then you can decide what to do in your own time.*

This research project has been approved by the University of Birmingham, University of Wales and Institute of Psychiatry, Kings College London (CREC/06/07-190) ethics committees.

Appendix 17

Information Pack sent to parents upon passing criteria

17a; Detailed information sheet.

17b; Consent form

17c; Demographic questionnaire pack

**Appendix 17a:
Information Sheet for Parents and Carers**

You and the person you care for are being asked to take part in a research study. Before you decide if you both wish to take part it is important that you understand why we are doing the research and what it will involve. Please take time to read the following information carefully and if it is appropriate, explain and discuss it with the person you care for. If there is anything that is unclear, or if you would like more information please contact us using the details provided at the end of the sheet.

What is research and what is the purpose?

This study is being conducted at the University of Birmingham in collaboration with the University of Wales, Bangor and the Institute of Psychiatry, Kings College London.

The research project is being conducted in association with the Cornelia de Lange Syndrome Foundation (UK & Ireland), Cri du Chat Syndrome Support group and the Angelman Syndrome Support, Research and Education Trust and is supported by the National Lottery, Community Fund.

The study aims to improve our understanding of the role of factors within the environment that may impact on self injurious and aggressive behaviours commonly associated with Cornelia de Lange, Cri du Chat and Angelman syndromes. The study will also examine the factors that are related to family well-being within these syndrome groups. We hope that greater understanding of the behaviours associated with these syndrome groups will help to support social inclusion, develop better intervention and behaviour management strategies and improve the health and well-being of affected individuals and their families.

Do we have to take part?

It is up to you and the person you care for whether or not you decide to take part, the decision you come to will not affect any services you receive, support from the syndrome groups or the availability of clinical consultations from Chris Oliver and his team at support group meetings. If you and the person you care for do decide to take part you will be asked to sign a consent form.

Will I be able to withdraw from the research?

Should you or the person you care for decide that you no longer wish to be involved in the research; you are free to withdraw your participation at anytime during the study and for a period of three months after the data collection with yourselves has been completed. If you decide to do so, information that you have provided in this time can also be withdrawn and destroyed without you giving reason. This will not restrict access to other services and will not affect the right to treatment.

Will our information be confidential?

All information collected will be kept on a confidential database that is only accessible to those working on the project. In the unlikely event of any evidence of abuse being identified, this information will be disclosed by the research workers. All personal details will be kept separately from the information collected and your child/person you care for will be identifiable by a code throughout the study to ensure anonymity. If published, information will be presented without reference to any identifying information.

What does it involve?

Participation in the research project will involve the following:

- You will be asked to complete two questionnaire packs for this study. The first questionnaire pack will provide us with general information about your child/person you care for and their abilities, it will also ask you (parent or legal guardian) for some information about yourself. The second questionnaire pack will ask questions regarding your child's/ person you care for's behaviour, sleep, communication and health. Finally, a third questionnaire pack will ask you questions concerning your wellbeing, and the impact that having a child with a genetic syndrome has on the family.
- We would like to take some time to discuss with you about your child's/person you care for's behaviour. Some of this will be done over the phone and some will be done during a home visit.
- We will visit your child/person you care for at their school, day centre or college 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 your child/person you care for's school, day centre or college 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 school environment. The first situation is a "high attention" in which the teacher or 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 teacher or 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 teacher or 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.

How video recordings will be made?

- Observations and video recordings will only take place during previously specified times that have been agreed by teachers and parents/ legal guardians.
- Video recordings will be kept and stored for further review by the Three Syndromes Project research team. When videotapes are not in use they will be stored in a locked cabinet in the School of Psychology, University of Birmingham and will only be viewed by research workers from the University of Birmingham. Information identifying your child will not be stored on or with the tape.
- Your child's privacy and dignity will be respected and video recordings will not take place if children are in a state of undress or when there is evidence that the observations are causing distress.
- Parents/ legal guardians and teachers can ask to see a copy of the videotape.
- The video recordings may only be viewed by legal guardians, individuals providing a service to the person, Professor Chris Oliver and research staff at the University of Birmingham. Any data that are derived from the tape will remain anonymous.
- We may contact you again in the future to ask your permission to use the video recordings for teaching purposes. At that stage you will be able to decide whether or not you are happy for the videos to be used for these purposes. However, agreeing to participate in this study does not mean that you will be obliged to give your permission for the use of these video recordings in the future.

At the end of the study

Each parent/ legal guardian will receive a personalised feedback report on their child/the person they care for.

A summary of the overall project's findings will be circulated to anyone involved who wishes to see a copy. Any requests for advice concerning your child/person you care for will be referred to Professor Chris Oliver, Clinical Psychologist. It is possible that you may be invited to participate in further research after the

study. However, consenting to participate in this study does not mean that you are obliged to do so.

Consent:

It is up to you whether or not you would like your child/person you care for to take part in the study. If your child/person you care for is aged between 2 and 15 years and you would like them to participate in this study please complete the enclosed consent form and return it to us in the envelope provided. If you feel it is appropriate, you may wish to discuss the project with your child/person you care for.

If you decide to become involved in the project then please complete the appropriate consent form and return this in the envelope provided. After you have returned your consent form you will be contacted by your allocated project worker who will give you further information about the project.

Who has reviewed the study?

This research project has been reviewed and approved by The University of Birmingham, School of Psychology Research Ethics Committee (0121 424 0594), the University of Bangor Ethics Committee (01248 382211 ext 8771) Kings College London Ethics Committee (0207 848 4020; ref: CREC/06/07-190)

Any concerns of queries?

If you are unclear about any aspect of the study or have any questions, please do not hesitate to contact Professor Chris Oliver by telephone: 0121 414 4909, by email: c.oliver@bham.ac.uk or at the following address:

Professor Chris Oliver
School of Psychology
University of Birmingham
Edgbaston
Birmingham, B15 2TT

Thank you very much for taking the time to read this information – please keep this information sheet for future reference

**Appendix 17b
Consent form**

Please initial the boxes

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

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

I consent to the processing of my personal information for the purposes of this research study. I understand that such information will be treated as strictly confidential and handled in accordance with the provisions of the Data Protection Act 1998.

I understand that as part of the above study, video/voice recordings of myself and my child/person I care for will be made and stored for further review.

I understand that the University of Birmingham will hold the copyright of any video/voice recordings collected during the study but that this does not entitle the University of Birmingham to edit, copy or use the videos for teaching purposes without my written permission.

I am happy to be contacted in the future by the University of Birmingham regarding the use of video recordings for teaching purposes.

I agree to participate in the above study.

I agree to the participation of my child / 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.....

Appendix 17c
Demographic Questionnaire pack

DEMOGRAPHIC QUESTIONNAIRE

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. I would be happy to be contacted for future research Yes No

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

1. Gender: Male Female

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

3. Is the person you care for verbal? (i.e. more than 30 signs/ words in their vocabulary)

Yes/ No (delete as appropriate)

4. Is the person you care for able to walk by themselves? Please tick where appropriate

1 = not at all 2 = not up stairs 3 = up stairs and elsewhere

5. Vision

1 = blind or almost 2 = poor 3 = normal

6. Hearing

1 = deaf or almost 2 = poor 3 = normal

7. Has the person you care for been diagnosed with a syndrome?

Yes/ No (delete as appropriate)

If yes, please indicate which syndrome in 5a. and answer questions 6 to 8. If no, please move on to question 9.

- 5a. Cornelia de Lange syndrome Cri du Chat syndrome
- Angelman syndrome

6. **What is the genetic mechanism causing the syndrome in the person you care for?**

- | | | | |
|---------------------|--------------------------|---------------------|--------------------------|
| Uni-parental disomy | <input type="checkbox"/> | Sequence repetition | <input type="checkbox"/> |
| Deletion | <input type="checkbox"/> | Translocation | <input type="checkbox"/> |
| Unknown | <input type="checkbox"/> | Other | |

7. **When was the person you care for diagnosed?**

8. **Who diagnosed the person you care for?**

- | | | | |
|---------------|--------------------------|---------------------|--------------------------|
| Paediatrician | <input type="checkbox"/> | Clinical Geneticist | <input type="checkbox"/> |
| GP | <input type="checkbox"/> | Other | |

9. **Has the person had any medical / health difficulties in the last six months? If yes, please give details:**

The following questions ask for background information about you, your child with Angelman Syndrome, and your family. Please tick the appropriate boxes or write in the spaces provided.

1. Are you male or female? Male Female

2. What was your age in years on your last birthday? _____years

3. Please tick the highest level of your educational qualifications.

- No formal educational qualifications.....
- Fewer than 5 GCSE's or O Level's (grades A-C), NVQ 1, or BTECH First Diploma.....
- 5 or more GCSE's or O Level's (grades A-C), NVQ 2, or equivalent.....
- 3 or more 'A' Levels, NVQ 3, BTECH National, or equivalent.....
- Polytechnic/University degree, NVQ 4, or equivalent.....
- Masters/ Doctoral degree, NVQ 5, or equivalent.....

4. What is your relationship to your child with Angelman Syndrome (e.g., mother, father, stepmother, grandmother, adoptive parent)? _____

5. In total how many people currently live in your home? _____ Adults _____ Children

6. Does your child with Angelman Syndrome normally live with you?

Yes No

If no, then where do they live? _____

7. What is your current marital status?

Married, and living with spouse.....

Living with partner.....

Divorced/Separated/Widowed/Single and NOT living with a partner....

If living with partner/spouse, please answer the following questions, if not, please go to question 12.

8. Is your partner male or female?

Male

Female

9. What was their age in years on their last birthday? _____ years

10. Please tick the highest level of your partner/spouses educational qualifications.

No formal educational qualifications.....

Fewer than 5 GCSE or O Level (grades A-C), NVQ 1, or BTECH First Diploma.....

5 or more GCSE or O Level (grades A-C), NVQ 2, or equivalent.....

3 or more 'A' Levels, NVQ 3, BTECH National, or equivalent.....

Polytechnic/University degree, NVQ 4, or equivalent.....

Masters/ Doctoral degree, NVQ 5, or equivalent.....

11. What is your partner/spouses relationship to your child with Angelman Syndrome (e.g., mother, father, stepmother, adoptive parent)?

12. Recent data from research with families of children with special needs has shown that a family's financial resources are important in understanding family member's views and experiences. With this in mind, we would be very grateful if you could answer the additional question below. We are not interested in exactly what your family income is,

but we would like to be able to look at whether those with high versus lower levels of financial resources have different experiences.

What is your current total annual family income? Please include a rough estimate of total salaries and other income (including benefits) before tax and national insurance/pensions.

Please tick one box only:

- Less than £15,000.....
- £15,001 to £25,000.....
- £25,001 to £35,000.....
- £35,001 to £45,000.....
- £45,001 to £55,000.....
- £55,001 to £65,000.....
- £65,001 or more.....

Appendix 18

Information to participants (Chapter 3)

18a: Cover letter to parents

18b: Information leaflet for parents and carers

18c: Consent form

Appendix 18a
Cover letter to parents

Ms. Gemma Griffith
School of Psychology
Bangor University
Brigantia Building
Penralt Road
Bangor,
Gwynedd
LL57 2AS

Dear X,

We are writing to ask for your help with the new research project. It is about parents/carers experiences of raising a child with (specific syndrome). We are particularly interested in what it has been like for you, as a parent, dealing with the issues that arose during your child's transition from childhood into adulthood. Specifically, how helpful (or unhelpful) social, educational, and medical services have been since your child reached adulthood. We hope do this by having an open-ended interview with you over the telephone so we can really get a sense of what it has been like for you, as a parent/carer.

This project is a part of the wider Three Syndromes project which you contacted us about earlier this year. Unfortunately X did not meet the criteria for that project due to their age. However, we know there is a lot to learn from the experiences of people like yourselves, and we would like to understand more about what parents/carers experiences during their child's transition from childhood into adulthood. This is an important area of study but has not yet been addressed within research.

This research is part of the Three Syndromes project which is being conducted at Bangor University in collaboration with the University of Birmingham and the Institute of Psychiatry, London. The research project is also being conducted in association with the Cornelia de Lange Syndrome Foundation (UK & Ireland), Cri du Chat Syndrome Support Group and the Angelman Syndrome Support, Research and Education trust and is supported by the Big Lottery Fund.

I enclose an information sheet that gives you some more detailed information about why the research is being carried out and what it will involve. Please take the time to read it, and if you are unclear about any aspect of the study or have any queries then please contact Gemma Griffith by telephone: 01248 388202, email psp017@bangor.ac.uk, or at the above address.

Yours sincerely,

Gemma Griffith



Appendix 18b
Information leaflet for parents and carers

We have selected you as a potential participant in a new study being conducted at Bangor University, in collaboration with the University of Birmingham and the Institute of Psychiatry, London. The study we are asking you to take part in has been developed following talks with parents and carers during data collection for the Three Syndromes project. Many parents have informally spoken to us about their experiences with medical, educational and social services and the impact of these services on their lives. Furthermore, it is common for parents to mention to us their concerns about what is going to happen to their child once they reach adulthood, as they feel unprepared and are not quite sure what to expect.

We would like to further explore this important issue with parents of adult children with (specific syndrome), as there is no research at all in this area, and little is known about how parents/carers like yourselves feel about adult services. It is hoped that the research will contribute towards gaining understanding of the kinds of issues that arise when dealing with social, educational, and medical services.

This may help parents of younger children with X syndrome to prepare for the childhood to adulthood transition, as well as being beneficial for families to know that there may be other families with adult children in similar circumstances. It may also inform social and medical services how to support the social inclusion of adults with rare genetic syndromes and their families.

What does it involve?

Participation in the research project will involve the following:

- If you are interested in taking part, please complete the enclosed consent form and return it in the pre-paid envelope.
- A researcher (Gemma Griffith) will phone you within two weeks of receiving the consent form to arrange a convenient time to conduct the phone interview.
- The interview will involve asking a few background questions, such as how many people live in your household, your adult child's abilities etc. We would then move on to talking about your experiences of dealing with the issues that have arisen during your child's transition from childhood into adulthood. Specifically, we would like to know what your experiences of social, educational, and medical services have been since your child reached adulthood. The interview will be recorded, with your permission, and is anticipated to last for around 45 minutes to an hour.

Consent:

If you decide to become involved in the project then you will be required to complete the enclosed consent form and return this to us.

Withdrawal:

Should you 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 any reason. Even after the interview has been completed, consent can be withdrawn and any data collected will be destroyed. This will not restrict your access to services and will not affect the right to treatment.

Confidentiality:

- When recordings of the interview are not being used they will be stored in a locked filing cabinet.
- Information identifying you or the child/person you care for will not be stored on or with the tape.

All information collected will be kept on a confidential database that is only accessible to those working on the project. If published, information will be presented without reference to any identifying information.

At the end of the study:

We will send you information on the results of the study. Any requests for advice concerning your child/person you care for will be referred to Professor Chris Oliver, Clinical Psychologist. It is possible that you may be invited to participate in further research after the study. However, consenting to participate in this study does not mean that you are obliged to do so.

Any concerns or queries?

If you are unclear about any aspect of the study or have any questions, please do not hesitate to contact Gemma Griffith by telephone: 01248 388202, by email: psp017@bangor.ac.uk or at address on the covering letter.

If you have any complaints about the way this research is being conducted you are welcome to address unresolved concerns to:

Dr. Oliver Turnbull
Head of School
School of Psychology
Bangor University
Brigantia Building
Penralt Road
Bangor,
Gwynedd
LL57 2AS

Appendix 18c
Consent form

Please initial the boxes

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

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

I agree to the interview being recorded.

I agree to participate in the above study.

Please complete the information below

Name..... Mr/Mrs/Miss/other (please circle)

Date of Birth.....

Name of your child..... Child's Date of Birth.....

Phone Number..... Best time of day to phone.....

Signature.....Date.....

Email address.....

FOR OFFICE USE ONLY

Signature of researcher.....Date.....

Appendix 19

Interview protocol**Prior to commencing interview session**

- Introduce yourself, check that it is still a good time to call
- Remind participants that the interview will be recorded, check that it is still ok to do this
- Remind participants of confidentiality and that any use of their data will not identify them as individuals.

Transition from childhood into adulthood- General, setting the scene questions.

- Tell me about X's transition into adulthood
- Was there any change in X as a person? Positive? Negative?
- Tell me about this
- Was/ is there anything that was particularly challenging for X during this time?
- Was/ is there anything that was particularly challenging for you during this time
- Has the relationship between you and X changed since they reached adulthood?
- (If relevant) What helped you cope during this time?
- Did you feel prepared for this transition?
- Did you know what to expect?
- Developmentally within your child
- From services?

- What was the transition away from school/college like for X?
- Was X prepared for the change?
- Did the school/college help X prepare for the change?
- How do you feel the transition process was handled by the school/college?
- Were you offered alternatives to school/collage or did you have to find out about these yourself?
- What were the alternatives? What did you think about these?
- Did you have any choice? If so, what made you choose a particular service?

Experiences with social services

- What kinds of services do you currently receive day-to day? Respite/ Day care/ College/ etc
- What do you think about these?
- Are there any current issues with any of the services?
- What is good about these services?
- Do you have any criticisms of these services?
- Ideally, what kinds of services would you like to have access to and how often?

Follow up questions on bad experiences

- How did this affect you?
- Did it affect your spouse/ family?

Specialist services

Has X needed specialist medical care since reaching adulthood?

- Tell me about it.
- What were the Doctors/nurses/specialists like?
- Did you feel involved in the process?
Do you feel that staff were adequately trained to meet X's and your needs?
- (if not clear) Was there anything positive about the experience?
- (if not clear) Was there anything negative about the experience?

Have you needed any specialist equipment for your home since X reached adulthood?

- What was the process of getting this like?

Has X needed any other forms of specialist assistance/input from specialised services?

- Tell me about this

Overview

- Overall, how would you evaluate the services X has received as an adult?
- What has been the main difference between adult and child services?
- (If relevant)- what effect has this change had on you?
- (If relevant) what effect has this change had on other family members?

Involvement

Do you feel you are listened to as a parent by services you have been in contact with?

- How does this make you feel?
- Are you as involved as you would like?
- Do you feel these services valued your input?

Rareness of syndrome

Ever encountered any problems due to the rareness of X's syndrome?

- What was this?
- Do you think it would make a difference if more people were aware of X syndrome?
- In what way?

Professionals

- How have you generally found the professionals you have come into contact with since x reached adulthood?

- Anything particularly good about anyone you have come across?
- (if yes) What qualities made them particularly good?
- Anything particularly bad about anyone you have come across?
- (if yes) What qualities did you find frustrating?

If not already discussed during the interview- ask these questions

- Has there ever been something you have found particularly good about any service or professional you have come across?
- Has there ever been something you have found particularly bad about any service or professional you have come across?

Future concerns

- Do you have any current thoughts about what the future might hold for X?
- Are you concerned about services for X in the future?
- Why/Why not?
- Looking back, what would have been helpful for you to know about the process of X transitioning into adulthood?
- What would your advice be to other parents of children with X syndrome approaching adulthood?

Additional filler questions may be required to expand some answers.

Examples are:

- You mentioned.....earlier, could you tell me more about this?
- We have talked about this already but is there anything you would like to add?

Appendix 20

Demographic information -Cross-syndrome study

1. Today's date: _____
2. Your name: _____

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

1. Gender: Male Female
2. Date of Birth: ___/___/___ Age: _____ years _____ months

The following questions ask for background information about you, and your family. Please tick the appropriate boxes or write in the spaces provided.

1. Are you male or female? Male Female

2. What was your age in years on your last birthday? _____ years

3. Please tick the highest level of your educational qualifications.

- No formal educational qualifications.....
- Fewer than 5 GCSE's or O Level's (grades A-C), NVQ 1, or BTECH First Diploma.....
- 5 or more GCSE's or O Level's (grades A-C), NVQ 2, or equivalent.....
- 3 or more 'A' Levels, NVQ 3, BTECH National, or equivalent.....
- Polytechnic/University degree, NVQ 4, or equivalent.....
- Masters/ Doctoral degree, NVQ 5, or equivalent.....

4. What is your relationship to your child with Angelman Syndrome (e.g., mother, father, stepmother, grandmother, adoptive parent)? _____

5. In total how many people currently live in your home? _____ Adults _____ Children

6. Does your child with Angelman Syndrome normally live with you?

Yes No

If no, then where do they live?

7. What is your current marital status?

- Married, and living with spouse.....
- Living with partner.....
- Divorced/Separated/Widowed/Single and NOT living with a partner....

If living with partner/spouse, please answer the following questions, if not, please go to question 12.

8. Is your partner male or female?

Male

Female

9. What was their age in years on their last birthday? _____ years

10. Please tick the highest level of your partner/spouses educational qualifications.

- No formal educational qualifications.....
- Fewer than 5 GCSE or O Level (grades A-C), NVQ 1, or BTECH First Diploma.....
- 5 or more GCSE or O Level (grades A-C), NVQ 2, or equivalent.....
- 3 or more 'A' Levels, NVQ 3, BTECH National, or equivalent.....
- Polytechnic/University degree, NVQ 4, or equivalent.....
- Masters/ Doctoral degree, NVQ 5, or equivalent.....

11. What is your partner/spouses relationship to your child with Angelman Syndrome (e.g., mother, father, stepmother, adoptive parent)?

12. Recent data from research with families of children with special needs has shown that a family's financial resources are important in understanding family member's views and experiences. With this in mind, we would be very grateful if you could answer the additional question below. We are not interested in exactly what your family income is, but we would like to be able to look at whether those with high versus lower levels of financial resources have different experiences.

What is your current total annual family income? Please include a rough estimate of total salaries and other income (including benefits) before tax and national insurance/pensions.

Please tick one box only:

- Less than £15,000.....
- £15,001 to £25,000.....
- £25,001 to £35,000.....
- £35,001 to £45,000.....
- £45,001 to £55,000.....
- £55,001 to £65,000.....
- £65,001 or more.....

Appendix 21

**Questionnaire on Resources and Stress Parenting and the family subscale
(QRS-F7)**

The following statements deal with your feelings about your child with disabilities. There are many blank spaces on the questionnaire (_____). Imagine the name of your child with disabilities in each of these blank spaces. Please give your honest feelings and opinions. Respond to all of the statements, even if they do not seem to apply. If it is difficult to decide "true" or "false", answer in terms of what you or your family do *most* of the time.

- | | | |
|--|------|-------|
| 1. Caring for _____ puts a strain on me | True | False |
| 2. Other members of the family have to do without things because of _____ | True | False |
| 3. In the future, our family's social life will suffer because of the increased responsibilities and financial stress. | True | False |
| 4. I can go to visit friends whenever I want _____ | True | False |
| 5. There are many places where we can enjoy ourselves as a family when _____ comes along | True | False |
| 6. Members of our family get to do the same kinds of things other families do _____ | True | False |
| 7. The constant demands to care for _____ limit my growth and development | True | False |

Appendix 22

Depression subscale of the Hospital Anxiety and Depression Scales

YOUR FEELINGS AND EMOTIONS

The following questions focus on how *you* feel about things. Please read each item and circle the reply underneath the item which comes closest to how you have been feeling in the past week. Do not take too long over your replies; your immediate reaction to each item will probably be more accurate than a long thought-out response.

- | | | | |
|--|---------------------------------------|-----------------------------------|----------------------------------|
| 1. I still enjoy the things I used to enjoy | | | |
| Definitely as much | Not quite so much | Only a little | Hardly at all |
| 2. I can laugh and see the funny side of things | | | |
| As much as I always could | Not quite so much now | Definitely not so much now | Not at all |
| 3 I feel cheerful | | | |
| Not at all | Not often | Sometimes | Most of the time |
| 4. I feel as if I am slowed down | | | |
| Nearly all the time | Very often | Sometimes | Not at all |
| 5. I have lost interest in my appearance | | | |
| Definitely | I don't take as much care as I should | I may not take quite as much care | I take just as much care as ever |
| 6. I look forward with enjoyment to things | | | |
| As much as I ever did | Rather less than I used to | Definitely less than I used to | Hardly at all |
| 7. I can enjoy a good book, radio or TV programme | | | |
| Often | Sometimes | Not often | Very seldom |

Appendix 23
Shortened version of the PAS (PAS5)

YOUR FEELINGS AND EMOTIONS

This scale consists of a number of words that describe different feelings and emotions. Read each item and then circle one of the responses. **Indicate to what extent you have felt this in the past week.**

	Very slight/ not at all	A little	Moderate	Quite a bit	Extremely
1. Enthusiastic	1	2	3	4	5
2. Alert	1	2	3	4	5
3. Inspired	1	2	3	4	5
4. Determined	1	2	3	4	5
5. Active	1	2	3	4	5

Appendix 24

The 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

Appendix 25
The Challenging Behaviour Questionnaire (CBQ)

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

Yes No

If the behaviour has not occurred, please go to question 6.

If the behaviour occurred in the past month please answer questions 2 to 5:

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

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

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

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

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

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

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

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

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

Yes No

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

Yes No

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

Yes No

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

Appendix 26

The Mood, Interest, and Pleasure Questionnaire

Instructions for completing the MIPQ-S

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

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

- | | | | | |
|---------------------------------------|--|--|--|-----------------------------|
| <i>interested all
of the time</i> | <i>interested
most
of the time</i> | <i>interested
about
half of the
time</i> | <i>interested some
of the time</i> | <i>never
interested</i> |
|---------------------------------------|--|--|--|-----------------------------|

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

- | | | | | |
|--------------------------------|---------------------------------|---------------------------------------|---------------------------------|------------------|
| <i>sad all of
the time</i> | <i>sad most
of the time</i> | <i>sad about half
of the time</i> | <i>sad some
of the time</i> | <i>never sad</i> |
|--------------------------------|---------------------------------|---------------------------------------|---------------------------------|------------------|

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*?

- | | | | | |
|----------------------------|---------------------------------|-----------------------------------|-----------------------------|--------------|
| <i>all of the
time</i> | <i>most of the
the time</i> | <i>about half of
the time</i> | <i>some of the
time</i> | <i>never</i> |
|----------------------------|---------------------------------|-----------------------------------|-----------------------------|--------------|

**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”*...

- | | | | | |
|----------------------------|---------------------------------|-----------------------------------|-----------------------------|--------------|
| <i>all of the
time</i> | <i>most of the
the time</i> | <i>about half of
the time</i> | <i>some of the
time</i> | <i>never</i> |
|----------------------------|---------------------------------|-----------------------------------|-----------------------------|--------------|

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

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

- | | | | | |
|----------------------------|-----------------------------------|--------------------------------------|--|---|
| <i>cried every
day</i> | <i>cried nearly
every day</i> | <i>cried 3-4 times
each week</i> | <i>cried once or
twice each
week</i> | <i>cried less than
once each
week</i> |
|----------------------------|-----------------------------------|--------------------------------------|--|---|

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

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

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

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

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

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

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

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

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

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

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

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

**facial expressions: interest might be indicated by the degree to which the person's gaze is being directed at the person/things involved in an activity.*

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

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

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

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

*gestures which appear to demonstrate enjoyment: e.g. clapping, waving hands in excitement etc.

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

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

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

*vocalizations: any words, noises or utterances.

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

Appendix 27

The Health Questionnaire

Instructions:

- Have these medical problems affected the person you care for in the past MONTH
- Please rate as **0** – if your child has not been affected by this problem in the past month, **1** - if they have been mildly affected, **2** – if the problem has moderately affected your child and **3** - if your child has been severely affected by the problem.

	No	Mild	Moderate	Sever.
1. Eye Problems (e.g. glaucoma / blocked tear duct/s).....	0	1	2	3
2. Ear Problems (e.g. infections, glue ear).....	0	1	2	3
3. Dental Problems (e.g. toothache / gum problems / mouth ulcers / delayed eruption of teeth).....	0	1	2	3
4. Cleft Palate.....	0	1	2	3
5. Gastrointestinal Difficulties (e.g. reflux / stomach problems).....	0	1	2	3
6. Bowel Problems (e.g. obstruction).....	0	1	2	3
7. Heart Abnormalities or Circulatory Problems (e.g. congenital heart lesions or murmur).....	0	1	2	3
8. Problems with Genitalia (e.g. prostate / testicular problems i.e. undescended testes)....	0	1	2	3
9. Hernia (e.g. inguinal or hiatal).....	0	1	2	3
10. Limb Abnormalities (e.g. malformed arm).....	0	1	2	3
11. Epilepsy / Seizures / Neurological Referrals.....	0	1	2	3
12. Lung or Respiratory Problems (asthma / bronchitis).....	0	1	2	3
13. Liver or Kidney Problems.....	0	1	2	3
14. Diabetes or Thyroid Function Problems.....	0	1	2	3
15. Skin Problems (e.g. tinea, eczema, psoriasis, dry skin).....	0	1	2	3
16. Other (please specify problem and severity from 0-3)	0	1	2	3

Examples of DVD-ROM content- introduction and prevalence of syndrome.
Understanding and Changing Challenging Behaviour in Angelman Syndrome



Written by: Chris Oliver, Jo Moss, Gemma Griffith, Jane Petty, Penny Tunnicliffe, Richard Hastings, Pat Howlin, Leah Bull, Darrelle Villa and Michael Yip

A Brief Introduction

On this DVD you will find information that will help you understand and change challenging behaviour in children and adults who have Angelman Syndrome. The information is derived from the results of the Three Syndromes research project that was funded by the Big Lottery. The information is organised in a way that will guide you through possible causes of challenging behaviours, how different causes can be assessed and how behaviours can be changed once you know the cause.

Appendix 28

**Examples of DVD-ROM content- introduction and prevalence of syndrome.
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When you are using the DVD you might feel a bit out of your depth and want to seek help and advice. If so, you can show or loan the DVD to professionals, such as GP's, clinical psychologists, Social Workers, teachers and others, so that they can help you with the process of understanding and changing challenging behaviours. On the DVD we have also included copies of research papers that can be downloaded and read by professionals.

If you are a professional then we hope that you will find the information and video clips helpful. The parents of the children featured in the video clips have given their permission for the clips to be included so that other parents and children might be helped. They have asked that these clips are not used in any other way, for example teaching or presentations, except by the Three Syndromes research team.

We are very grateful to the Big Lottery who funded the research project and to the Three Syndrome support groups who were involved in the research. The Cornelia de Lange Syndrome Foundation (UK and Ireland), the Cri du Chat Syndrome Support Group and the Angelman Syndrome Support Education and Research Trust worked very hard to ensure the project was successful and were active partners at each stage of the research. Finally, we are very grateful to the parents and children who did not hesitate to take part in the research and in doing so made this DVD possible.

Prof. Chris Oliver
Cerebra Centre for Neurodevelopmental Disorders
School of Psychology
University of Birmingham
www.cndd.bham.ac.uk

Prevalence of Angelman Syndrome

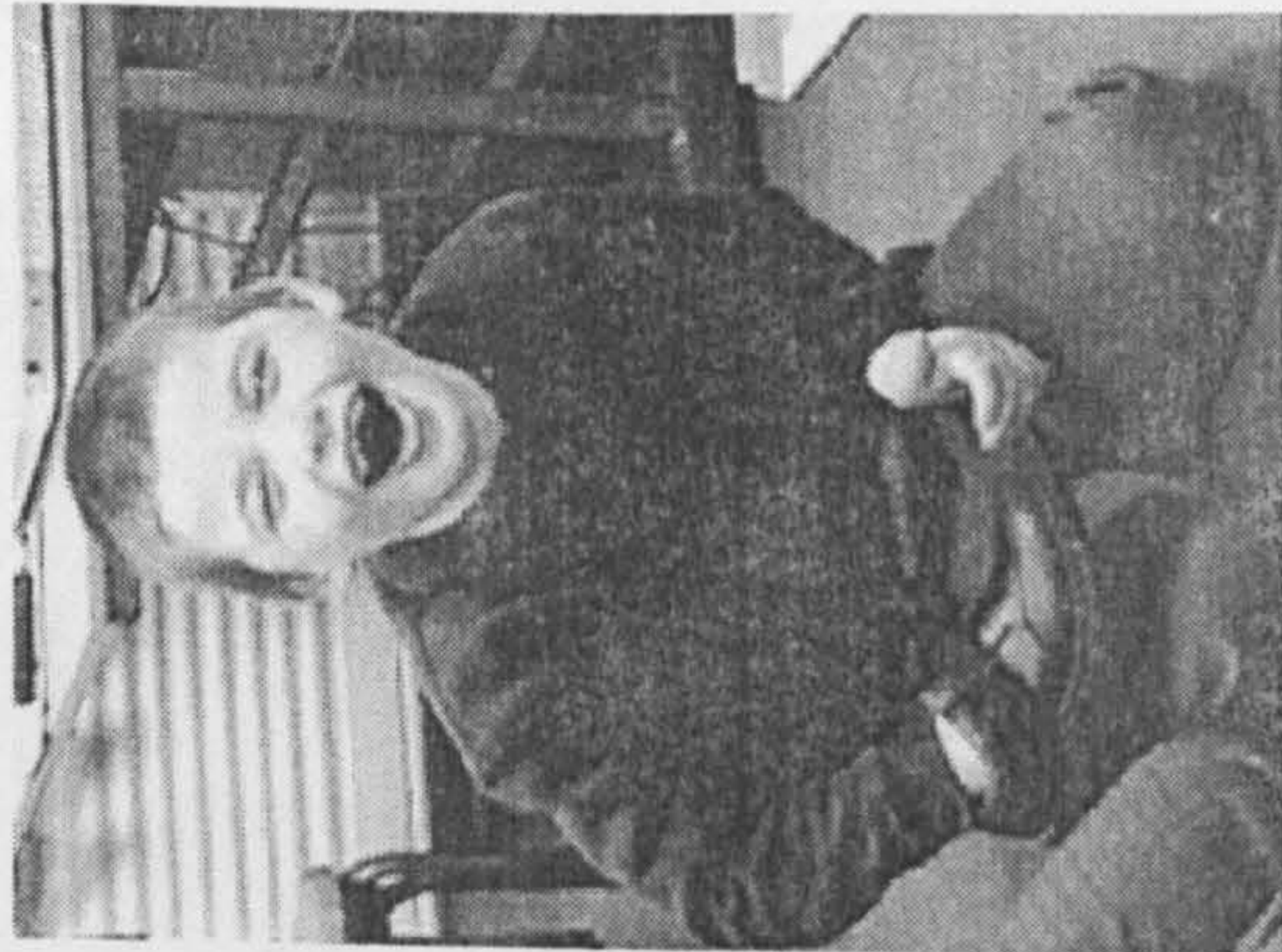
'It is important to remember that a person is a person first, and the diagnosis is secondary'

Angelman Syndrome is a rare genetic disorder and at present the prevalence has been estimated to be between 1 in 10 000 and 1 in 40 000 live births.

Angelman Syndrome is named after Dr Harry Angelman who first described three children with the syndrome in 1965. After working with the children he was inspired to write an article after seeing a painting in a museum in Verona, Italy entitled "A Boy with a Puppet". This painting gave him the idea of writing a paper titled "Puppet Children".



In his writing, Dr Angelman described the children as "Happy Puppet Children" because of their happy character and stiff jerky movements and this term was used widely until the late eighties. In 1982, two researchers (Williams and Frias) considered the term 'Happy Puppet' to be impolite and so the name of the disorder was changed to 'Angelman Syndrome'.



'I'd say she's probably both the best and the worst thing that's ever happened to me. The worst obviously when you find out that your child is going to be disabled but the best in that she's brought out so many different sides and so much understanding of other people'