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Psychological well-being in parents of children with Angelman, Cornelia de Lange and Cri du Chat syndromes

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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, Cornelia de Lange syndrome and Cri du Chat syndrome.
Method Parents of children with Angelman syndrome ($n = 15$), Cornelia de Lange syndrome ($n = 16$) and Cri du Chat syndrome ($n = 18$), and a matched comparison group of parents of children with autism and intellectual disabilities ($n = 20$) completed questionnaires on both psychological distress (stress, anxiety, depression) and positive psychological functioning.
Results Parents of children with Angelman syndrome consistently reported the highest levels of psychological distress, and parents of children with Cornelia de Lange syndrome the lowest, with parents of children with Cri du Chat syndrome and autism scoring between these two. Positive psychological functioning was 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.

Keywords Angelman syndrome, Cornelia de Lange syndrome, Cri du Chat syndrome, parents, stress

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Introduction

Advancements in genetic 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 ID, such as autism and Down syndrome (e.g. Hodapp 1997; Sanders & Morgan 1997; Olsson & Hwang 2001; 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.

Angelman syndrome is estimated to occur in between 1:10 000 to 1:40 000 live births (Clayton-Smith 1993; Buckley *et al.* 1998). The severity of ID in Angelman syndrome ranges from moderate to profound (Clarke & Marston 2000). Common behavioural features of the syndrome include frequent smiling and laughing, hyperactivity and sleep disorder (Clayton-Smith & Laan 2003; Horsler & Oliver 2006).

It is estimated that Cornelia de Lange syndrome occurs in around 1:40 000 live births (Beck 1976; Beck & Fenger 1985). The majority of individuals with Cornelia de Lange syndrome have profound or severe ID (Berney *et al.* 1999). Common behavioural features include: anxiety, oversensitivity, sensory self-stimulation, self-injurious behaviour and compulsivity (Basile *et al.* 2007).

Cri du Chat syndrome is so named because of a characteristic 'cat-like' cry, apparent immediately after birth. Estimated prevalence is between 1:37 000 and 1:50 000 live births (Niebuhr 1978; Higurashi *et al.* 1990). The degree of ID ranges from profound to moderate, and behavioural patterns associated with the syndrome include self-injurious behaviour and hyperactivity (Cornish *et al.* 1998; Cornish & Bramble 2002; Sarimski 2003).

These three syndromes are of interest 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 (Kasari & Sigman 1997; Baxter *et al.* 2000; Hastings *et al.* 2005a; Most *et al.* 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.

In the only study on families of children with Angelman syndrome ($n = 22$), Van den Bourne *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 similarly high for both groups of parents.

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 (Abidin 1990). Richman *et al.* (2009) 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 Parenting Stress Index. Parental stress was associated with high levels of child self-injury, stereotypy, and lower levels of child prosocial and adaptive behaviour.

Hodapp *et al.* (1997a) 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.

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 Angelman, Cornelia de Lange and Cri du Chat syndromes, which explored child behaviour problems and the association with parental stress (Hodapp *et al.* 1997a; Richman *et al.* 2009; 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 *et al.* 2001). Although evidence is equivocal as to whether mothers and fathers react differently to raising a child with an ID (e.g. Van den Bourne *et al.* 1999; McCarthy *et al.* 2006; Shin *et al.* 2006), 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 *et al.* 2010).

To what extent 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 between parental outcomes and issues arising from the rarity of a syndrome 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 *et al.* 1997b; 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 ID. 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. Hastings & Taunt 2002; Blacher & Baker 2006). 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 ID 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 ID as an appropriate comparison group. Prevalence rates for autism vary from 13–34 per 10 000 individuals (Fombonne 2003; Yeargin-Allsopp *et al.* 2003) to more recent figures of 110 per 10 000 individuals (Kogan *et al.* 2009). It is further estimated that around 68% of individuals with autism also have a cognitive impairment (Yeargin-Allsopp *et al.* 2003). The decision to include children with autism and ID as a comparison group 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 ID 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; Duarte *et al.* 2005; Blacher & McIntyre 2006; Herring *et al.* 2006; Lewis *et al.* 2006; Mugno *et al.* 2007; Rutgers *et al.* 2007; Schieve *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 explore well-being among parents of children and adolescents with Angelman, Cornelia de Lange and Cri du Chat syndromes. The authors also aimed to address the five methodological issues identified above, by comparing positive and negative well-being of both mothers and fathers of children 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 ID 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

Sixty families (20 from each rare syndrome group) were recruited for a wider study about the three rare syndromes. 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. Criteria Number 3 was necessary as the wider study involved observations of challenging behaviour, thus requiring a reasonably high frequency of presenting behaviour.

The data from parents with a child with autism in the present study were taken from a larger study of families of children with an ID (Hastings *et al.* 2005b). Autism diagnosis was based on parental report of a formal diagnosis. 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 (Rojhan *et al.* 2001). Children who were rated as engaging in any aggressive or self-injurious behaviour either daily or hourly on the frequency scale of the Behavior Problems Inventory were eligible for inclusion. Second, children had a Vineland Adaptive Behavior Scale (VABS) adaptive behaviour composite score of <70. This process resulted in the selection of 20 families whose child with autism met both criteria.

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 ID (20 mothers and 7 fathers). Demographic details for all four aetiology groups are summarised in Table 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). Previous studies examining adjustment among parents of children with Cornelia de Lange syndrome make no reference to whether the parents were the biological or adoptive parents (Sarimski 1997; Richman *et al.* 2009; Wulffaert *et al.* 2009). It is therefore unknown whether these are usual adoption rates among children with Cornelia de Lange syndrome or whether it is an artefact of the current sample. Independent *t*-tests were conducted and no significant differences emerged on outcome variables between biological mothers and adoptive/foster parents. Recent longitudinal research suggests that parental adjustment does not markedly differ between biological families and adoptive families of children with ID (Glidden & Jobe 2009).

A series of one-way between-subjects ANOVAs and chi-squared tests were conducted on demographic variables across the four groups. Significant group effects were found on maternal age ($F_{3,63} = 6.39$, $P \leq 0.01$) and on the VABS (Sparrow *et al.* 1984, 2005) adaptive behaviour composite ($F_{3,65} = 11.41$, $P \leq 0.001$). *Post hoc* tests were then used to explore

Table 1 Demographic information on the four aetiology groups with *post hoc* analysis

Demographics	Angelman syndrome (AS; *n* = 15)	Cornelia de Lange syndrome (CdLS; *n* = 16)	Cri du Chat syndrome (CdCS; *n* = 18)	Autism (*n* = 20)	*Post hoc* test
Child age range (years)	3.0–18.7	5.0–18.6	2.2–16.0	3.7–15.2	–
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	10	4	16	
CBI total number of challenging behaviours: mean (SD)	6.47 (2.53)	4.64 (1.28)	5.89 (1.91)	n/a	–
VABS composite score	41.13 (10.68)	39.44 (10.69)	53.44 (7.18)	38.00 (7.16)	CdCS > CdLS**, AS**, Autism**
Maternal age: range and mean (SD) (years)	32–50 41.79 (6.04)	37–65 47.31 (8.90)	31–50 39.56 (5.22)	27–47 37.89 (6.18)	CdLS > CdCS**, Autism**
Paternal age: range and mean (SD) (years)	32–48 42.38 (4.82)	30–65 47.60 (10.38)	31–48 41.92 (4.92)	28–47 39.14 (6.41)	–
% of primary caregivers married or living with a partner	(13/15)† 87.6	(16/16) 93.6	(18/18) 66.6	(17/20) 70	–
% of families earning below £25 000	(14/15) 28.6	(15/16) 33.3	(17/18) 47.1	(15/20) 75	–

* $P < 0.05$; ** $P < 0.01$.

† The figure shows total number of parents who responded to the question.

CBI, Challenging Behaviour Interview; VABS, Vineland Adaptive Behavior Scale.

pairwise differences and these are displayed in Table 1.

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 VABS 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: Socialisation, Daily Living Skills, Communication and Motor Skills (used for children under 7 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 (VABS-II; Sparrow *et al.* 2005), which measures the same four domains as the earlier version of the VABS. The data set 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 0.80 to 0.95, and inter-rater reliability, with correlation coefficients from 0.75 to 0.85 (Sparrow *et al.* 2005). The VABS demonstrates good convergent validity with the VABS-II, with most of the adjusted correlations between sub-domains being in the upper 0.80s and 0.90s (Sparrow *et al.* 2005). Administering the VABS over the telephone has been used successfully in previous research (Hastings *et al.* 2005a; Philofsky *et al.* 2007). Conducting the VABS over the telephone has been found to be a valid method of administration, and is highly correlated with conducting the VABS using face-to-face interviews (Limperopoulos *et al.* 2006).

The Challenging Behaviour Interview (CBI; Oliver *et al.* 2003) was used to determine the total number of challenging behaviours exhibited by the child within the last month. Parents were asked to rate how many out of 13 behaviours (e.g. self-injury, inappropriate vocalisations, aggressive behaviour, stereotyped behaviour, smearing) had occurred within the last month. Each behaviour type

is presented to the parent along with a fully operationalised definition. For example, self-injury is defined as 'non-accidental behaviours which produce temporary marks or reddening of the skin or cause bruising, bleeding or other temporary or permanent damage'. The interview was conducted face-to-face with parents of the child. The CBI has good mean kappa indices for inter-rater reliability (0.67) and test–retest reliability (0.86; Oliver *et al.* 2003).

In addition, parents of children in the three rare syndrome groups completed five questionnaire scales. The Parent and Family Problems sub-scale from the Questionnaire on Resources and Stress – Short Form (QRS-F; Fredrich *et al.* 1983) was used to measure general parental stress related to having a child with a disability. Five items were excluded from the original sub-scale 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 0.78, and for fathers 0.89.

The Hospital Anxiety and Depression Scales (HADS) were used to assess parental mental health (Zigmond & Snaith 1983). 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 *et al.* 2004). The HADS contains 14 4-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 *et al.* 2005b). Cronbach's alpha for the present sample of mothers of children with rare genetic syndromes was 0.88, and for fathers 0.91.

The Positive Affect Scale was derived by extracting the 10 positive affect items from the Positive and Negative Affect Scale (Watson *et al.* 1988). Parents were asked to rate to what extent the 10 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 0.91 and 0.92 for mothers and fathers respectively.

The Positive Gain Scale (Pit-ten Cate 2003; MacDonald *et al.* 2010) 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 0.71 and 0.75 respectively.

The final questionnaire measure was devised for the purposes of the present research. The Genetic Syndrome Stressors Scale (GSSS) 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 ID were reviewed. Second, semi-structured telephone interviews were conducted with six parents of children with Angelman, Cri du Chat or Cornelia de Lange syndromes (none of the parents interviewed participated in the current study). 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. Items include 'Not having access to professionals who have knowledge about my child's condition' and 'Having to explain my child's condition to new people I meet'. The resulting questionnaire had 14 items. Based on a total score across all 14 items, Cronbach's alpha for the current sample was 0.83 and 0.87 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

$r = 0.59$), depression ($r = 0.55$) and stress ($r = 0.61$), and negatively correlated with Positive Affect ($r = 0.41$). For fathers, the GSSS was positively correlated with anxiety ($r = 0.47$), depression ($r = 0.52$) and stress ($r = 0.46$), and negatively correlated with Positive Affect ($r = 0.33$). All the above correlations had significance values of $P < 0.05$. These data suggest that the GSSS has good face validity and internal consistency, and the correlation data suggest moderately good concurrent validity.

Procedure

This study was part of a wider project investigating the behavioural functioning of children with the three rare syndromes as well as the current family adjustment research.

A database held by the research team contained information about 127 individuals with Angelman syndrome, 68 individuals with Cri du Chat syndrome and 157 individuals with Cornelia de Lange syndrome (ages ranged 1–48 years). All families on the database who had previously given consent for being contacted about future research, and whose child met the criteria of the study, were mailed a letter and an information leaflet explaining the nature of the research and the inclusion criteria ($n = 31$). Of these families, 21 returned consent. A further 23 families were recruited through mailing flyers to families via national parent syndrome support groups, and 16 families were recruited via announcements at syndrome support group family conferences throughout the UK.

If parents expressed interest in participating by contacting the research team, a researcher would then make telephone contact within 7 days to determine whether potential participants currently met the three inclusion criteria for the study. The Challenging Behaviour Questionnaire (Hyman *et al.* 2002) 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, he or she was included in the current study.

Following screening, parents were mailed a consent form, a detailed information sheet about the wider study and a demographic questionnaire pack. Once consent was received, the family questionnaire packs were mailed, and the VABS-II was conducted via the telephone with the main caregiver within 2 weeks of the questionnaire pack being sent. Families were followed up by telephone if the questionnaire packs had not been returned within 4 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 because of 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 because either of divorce or of 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 parents of the children with autism and an ID completed the HADS, the QRS-F Parent and Family Problems sub-scale, and the Positive Affect Scale via postal questionnaire. The Positive Gain Scale, the GSSS and the CBI 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 and depression scale of the HADS, as recommended by Zigmond & Snaith (1983), a higher percentage of parents in this study had likely clinical levels of symptoms compared with normative UK data (Crawford *et al.* 2001; see Table 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%). The assumptions for chi-squared tests were not met because of the small sample size, 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.*

Table 2 The number of mothers and fathers at or above clinical cut-off levels for anxiety and depression

	Number of mothers/women reaching clinical cut-off		Number of fathers/men reaching clinical cut-off	
	Anxiety (%)	**Depression (%)**	**Anxiety (%)**	**Depression (%)**
Angelman syndrome	10/14* (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† (%)	12	4	6	2

* These figures indicate how many out of the total number of mothers or fathers reach clinical cut-off scores.
† Normative scores are based on Crawford *et al.* (2001).
ID, intellectual disability.

2001). The observed distributions differed significantly from the normative distribution for both mothers and fathers on anxiety and depression ($P < 0.05$) 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.

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 ($P \leq 0.01$) and Cornelia de Lange syndrome ($P \leq 0.01$). Mothers of children with autism were significantly more likely to report clinical levels of anxiety than mothers of children with Cornelia de Lange syndrome ($P \leq 0.05$). 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 ($P \leq 0.05$). The likelihood of reporting clinical levels of depression was significantly greater for fathers of children with Angelman syndrome than for fathers of children with autism ($P \leq 0.01$) and Cornelia de Lange syndrome ($P \leq 0.001$). Fathers of children with Cri du Chat syndrome were also more likely to report clinical levels of depression than fathers of children with autism ($P \leq 0.001$) and Cornelia de Lange syndrome ($P \leq 0.001$).

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 ($F_{3,62} = 5.61$, $P \leq 0.01$) and paternal ($F_{3,41} = 6.34$, $P \leq 0.01$) ratings of parental stress. When a significant group effect was found, *post hoc* Tukey's tests were used to examine pairwise differences, and these are summarised in Table 3. Where there were statistically significant group effects (see Table 1), 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) because of 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 Corne-

Table 3 Maternal and paternal scores for the four aetiology groups

	Maternal outcomes: mean (SD)					Paternal outcomes: mean (SD)				
Measure	**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*, CdCS*	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)	–	–

* $P < 0.05$; ** $P < 0.01$.

AS, Angelman syndrome; CdLS, Cornelia de Lange syndrome; CdCS, Cri du Chat syndrome; HADS, Hospital Anxiety and Depression Scale; QRS-F, Questionnaire on Resources and Stress – Short Form; GSSS, Genetic Syndrome Stressors Scale.

lia 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 ($F_{3,61} = 5.61$, $P \leq 0.01$), 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 effect for paternal stress ($F_{3,41} = 6.34$, $P \leq 0.01$), 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 affect when examining positive outcomes.

Table 4 demonstrates the effect sizes on maternal and paternal measures between the three rare syndromes. Significant differences between the syndrome groups are accompanied by high effect sizes, again suggesting a robust difference between rare syndrome groups.

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 with 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

Table 4 Maternal and paternal between-group effect sizes (Cohen's *d*)

	Maternal outcomes			Paternal outcomes		
Measure	**AS vs. CdLS**	**AS vs. CdCS**	**CdCS vs. CdLS**	**AS vs. CdLS**	**AS vs. CdCS**	**CdCS vs. CdLS**
HADS anxiety	0.64	0.64	0.14	1.00	0.30	0.69
HADS depression	0.30	0.37	0.01	0.97	0.33	0.76
QRS-F Family Problems sub-scale	1.67**	1.24*	0.49	2.02**	0.81	0.71
Positive Affect Scale	0.32	0.05	0.36	0.56	0.01	0.63
Positive Gain Scale	0.26	0.15	0.40	0.91	0.01	0.73
GSSS	0.49	0.50	0.07	0.50	0.49	0.07

* $P < 0.05$; ** $P < 0.01$.

AS, Angelman syndrome; CdLS, Cornelia de Lange syndrome; CdCS, Cri du Chat syndrome; HADS, Hospital Anxiety and Depression Scale; QRS-F, Questionnaire on Resources and Stress – Short Form; GSSS, Genetic Syndrome Stressors Scale.

and fathers of children with Angelman syndrome had the highest levels of negative outcomes even than a comparison group of parents of children with autism (albeit this difference reached significance on the QRS-F Parent and Family sub-scale only); and (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 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. Small sample sizes are a common difficulty within research on rare syndromes, and efforts to recruit larger samples would be useful in future research. The mean scores and effect sizes in Tables 3 and 4 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 ID.

Secondly, the children included in this study all displayed at least one episode of challenging behaviour a day, and thus represent a subset of children with these rare genetic syndromes who have higher frequencies of behaviour problems. This needs to be borne in mind when interpreting the results from this study, as they may pertain only to this particular subgroup of families whose children display frequent challenging behaviour. However, the little research that has been conducted on parents of children with Angelman, Cri du Chat and Cornelia de Lange syndromes (without selecting for challenging behaviour) also consistently suggests that parental stress levels are generally high among these populations (Hodapp *et al.* 1997a; Sarimski 1997; Van den Bourne *et al.* 1999; Richman *et al.* 2009; Wulffaert *et al.* 2009). Further research is needed to help examine this complex issue.

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.

Finally, parents scored within the mid-range of scores on the GSSS, with similar patterns between the groups as found by well-validated measures such as the QRS-F (Fredrich *et al.* 1983). However, the psychometrics of the GSSS are still relatively unknown and thus the results should be interpreted with caution.

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? The high stress levels in parents may be because of a combination of high levels of challenging behaviour and other 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), which 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, as 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 parents 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 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 ID 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 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.* 2009), 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. Similarly, a replication of this study using families of children with these rare syndromes without an inclusion criterion involving high frequencies of challenging behaviour may be informative.

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 and high frequencies of challenging behaviour, even when compared to parents of children with autism. The results from this study and previous research (Richman *et al.* 2009; Wulffaert *et al.* 2009) suggest that access to appropriate intervention is essential for parents of these children. 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, which may help to alleviate parental guilt (Hodapp *et al.* 1997b). Finally, there is also evidence that the provision of parental workshops targeting parents' own cognitions about their child may help to reduce parental stress (Shin *et al.* 2006).

Second, as the prevalence of challenging behaviours is high among these three aetiology groups (Hodapp *et al.* 2003; Horsler & Oliver 2006; Richman *et al.* 2009), 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).

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