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

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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).

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.

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 outcomes were similar across the four aetiology groups.

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

KEY WORDS: Angelman syndrome, Cornelia de Lange syndrome, Cri du Chat syndrome, parental, stress, positive perceptions.
Introduction

Advancements in genetics research have led to a growing interest in describing the behavioural phenotype 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 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). We found five studies in total 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, ‘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). Unlike the two later studies on Cornelia de Lange syndrome, 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 had stress levels that reached the Parenting Stress Index’s (PSI) cut-off point for ‘very high’ (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 predictor of increased parental stress, but was not as strongly associated as child problem behaviours.
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 by addressing five methodological issues: (1) Child Behaviour Problems as a confounding factor, (2) Mother-father differences, (3) Stresses associated with the rareness of the syndrome, (4) Positive as well as negative parental outcomes, and (5) Autism as a benchmarking condition for parental distress. Each of these issues is discussed briefly below.

The first methodological issue is that in all three family studies of these syndromes which explored child behaviour problems, found that they were to be a predictor of parental stress (Hodapp et al., 1997; Richman et al., in press; Wulffaert et al., 2009). However, it is not known whether the samples included children who showed no behaviour problems at all since this was not an explicit inclusion criterion in these studies. Therefore, it is difficult to evaluate whether any family outcome differences between syndromes are an artefact of recruitment procedures in relation to child behaviour problems. In the present study, we recruited only families of
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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 ID (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 issues pertaining uniquely to the rareness of the child’s syndrome affect family experiences is the third methodological point. This is a question seldom explored within the family literature on rare genetic syndromes. Where associations with rareness 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). Researchers have neglected 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).

The fourth methodological issue is that there has been gathering interest in the putative positive impact of having a child with ID in the family. Existing research 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 explored positive as well as negative psychological well-being.

Finally, there is a difficulty in benchmarking the 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). We would like to propose families of children with autism and ID as a sensible benchmark group. The reason for this proposal is that parents of children with autism reliably report more psychological distress than parents of typically developing children, parents of children with 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; Blacher & McIntyre, 2006; Duarte, Bordin, Yazigi, & Mooney, 2005; Herring et al., 2006; Lewis et al., 2006; Mugno, Ruta, D’Arrigo, & Mazzone, 2007; Scheive, Blumberg, Rice, Visser, & Boyle, 2007; Rutgers et al., 2007).

To address these five methodological points in the present research, we compared 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 ID to help benchmark the 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 ID (20 mothers and 7 fathers). The data from 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 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. When a significant group effect was found, post-hoc tests were used to explore 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 composite score) on the Vineland Adaptive Behavior Scales (VABS: Sparrow, Balla, & Cicchetti, 1984) or VABS- Second edition (VABS II: Sparrow, Cicchetti, & Balla, 2005).
Measures

The VABS (Sparrow et al., 1984; Sparrow et al., 2005) 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 can be 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 II (Sparrow et al., 2005), which measures the same four domains as the earlier version of the VABS. The original 1984 version of the VABS was used for the autism and ID group. The VABS-II also 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 to a questionnaire to assess demographic variables, 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) was used to measure general parental stress related to having a child with disability in the family. 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). 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). 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) 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) is a measure 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 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. 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 (see Appendix). 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.

Data from the present study allowed us to carry out a preliminary exploration of the concurrent validity of the GSSS. Pearson’s correlations showed that the GSSS was moderately positively associated with maternal anxiety ($r = .59$), 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 ($r = -.33$). The present data suggest that the GSSS may have 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 were recruited for the wider study on child behaviour problems with 20 families from each rare syndrome group. All children: 1. Had a 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 an information leaflet explaining the nature of the research and the inclusion criteria. 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 Interview (CBI: Oliver et al., 2003) 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 that were 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 national parent syndrome support groups, and were also screened using the same procedure. 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 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 being mailed.

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 at least one parent did complete a questionnaire pack but their partner did not, this is 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) in the present study were taken from a larger study of families of children with ID (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). 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 ID 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 either 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 2). The most striking result is that a much higher percentage (71.4%) of mothers of children with Angelman syndrome were 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). It was found that the observed distributions significantly differed from the normative distribution for both mothers and fathers on anxiety and depression (p<.05) in all four aetiology groups. The one exception was fathers of children with Cornelia de Lange syndrome, who did not significantly differ 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 significantly 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 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). The same was found for fathers of children with Cri du Chat syndrome, who 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).

----------------------Insert Table 2 about here-------------------------

Group differences for maternal and paternal outcomes

Between-group ANOVAs were used to explore maternal and paternal data across the syndrome groups and the autism group. 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. Where there were statistically significant group effects, the analyses were repeated including maternal age, child age, and child ABC 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, with the exception of the analysis of father depression scores. There was a statistically significant group effect for paternal depression scores in the ANOVA (F(3, 41) = 2.66, p=.048) but not when using a non-parametric test (p= .108).
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 and autism being in between the two other groups. 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 due to mothers of children with Angelman syndrome reporting significantly higher stress levels than parents of children with Cornelia de Lange syndrome, Cri du Chat, and autism. The positive impact of the child on the family and maternal positive affect did not differ significantly between groups.

The results from paternal outcomes followed the same trend as maternal outcomes, although the mean scores for fathers were lower than for mothers. 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 and autism being in between the two. 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 due to fathers of children with Angelman syndrome reporting significantly higher levels than parents of children with Cornelia de Lange syndrome. Although the group effect for father depression was not significant ($F(3, 41) = 2.66, p=.061$), post-hoc analysis revealed that fathers of children with Angelman syndrome reported significantly higher levels of depression than fathers of children with Cornelia de Lange syndrome ($p=.048$).

There was no significant group effect when examining positive outcomes, although the mean scores reveal that fathers of children with Cornelia de Lange
syndrome rated their child as giving more positive contributions, and reported higher positive affect than fathers of children with Cri du Chat or Angelman syndrome.

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, and 4. There were no consistent group differences for parental positive well-being outcomes. This pattern of results was found even after the groups were selected for the frequency of behaviour problems that have been found in previous research 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) and this will have reduced the power of any statistical tests 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 indicate that there may well be further meaningful group differences that
could emerge given larger samples in future research. The general pattern of the group means reflects the main finding from this research 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 ID. 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. Therefore, these participants may represent a particularly well-informed and committed group of parents, and thus the representativeness of the samples is unknown.

Is there something about the behavioural phenotype of children with Angelman syndrome that contributes to highly elevated stress and anxiety levels in parents? There may be a hitherto unexplored aspect of Angelman syndrome that may explain this finding. 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 such as laughing and smiling behaviours associated with Angelman syndrome are hypothesised to increase attention from mothers (Oliver et al., 2007). Although naturally perceived as a positive attribute, could increased sociability 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 draining for parents. Perhaps focusing on aspects of the
behavioural phenotype such as sociability will help unravel any aetiology-related
causes of the increased parental stress found among parent of children with
Angelman syndrome.

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, indicating that many parents experienced stressors that may be related to the
rarity of their child’s syndrome. Furthermore, scores on the GSSS were moderately
correlated with both negative and positive well-being measures. These data suggest
that stress associated with rarity of syndromes may be worthy of additional research
attention in future, including qualitative research studies that may 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. Our 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 as 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 this
is the first research study to separate out father well-being in this population it is difficult to ascertain why they are reporting less stress than all other groups. As the group of fathers was rather small (n=14) replication of this research is needed to see whether the current findings are a real reflection of how fathers of children with Cornelia de Lange syndrome adapt to their family situation.

Even given the studies limitations, the observed syndrome differences demonstrate the high degree of stress and the vulnerability to experiencing clinical levels of anxiety and depression that parents of children with these rare genetic syndromes undergo, even when compared to parents of children with autism. The results from this study and previous research (Richman et al., in press; Wulffäert et al., 2009) suggest that parents of these three rare syndromes need to be made priority groups in the receipt of any interventions. Intervention strategies for parents could thus be twofold. As the prevalence of challenging behaviours is high among children with these syndromes, early behavioural interventions could be implemented with the aim of 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. A 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 which take into account the genetic syndrome of the child could emphasise that some types of problem behaviours among children with rare syndromes are genetically determined, which may help reduce parental guilt
(Hodapp, 1997). Interventions surrounding parental cognitions may also be useful. There is evidence to suggest that parental workshops targeting parent’s own cognitions about their child may help to reduce parental stress (Singh et al., 2006)
References


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Appendix

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?

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

<table>
<thead>
<tr>
<th>Demographics</th>
<th>Angelman syndrome (n =15)</th>
<th>Cornelia de Lange syndrome (n =16)</th>
<th>Cri du Chat syndrome (n = 18)</th>
<th>Autism (n =20)</th>
<th>Post hoc test</th>
</tr>
</thead>
<tbody>
<tr>
<td>Child age range</td>
<td>3.0 -18.7 years</td>
<td>5.0-18.6 years</td>
<td>2.2-16.0 years</td>
<td>3.7-15.2 years</td>
<td>--</td>
</tr>
<tr>
<td>Child age (Mean, SD)</td>
<td>10.07 (4.79)</td>
<td>11.75 (3.49)</td>
<td>7.83 (4.66)</td>
<td>9.30 (3.37)</td>
<td>CdLS&gt; CdC*</td>
</tr>
<tr>
<td>Child gender (n males)</td>
<td>10 males</td>
<td>10 males</td>
<td>4 males</td>
<td>16 males</td>
<td></td>
</tr>
<tr>
<td>VABS composite score</td>
<td>41.13 (10.68)</td>
<td>39.44 (10.69)</td>
<td>53.44 (7.18)</td>
<td>38.00 (7.16)</td>
<td>CdC&gt;CdLS**, AS**, Autism**</td>
</tr>
<tr>
<td>Maternal age, range and mean (SD)</td>
<td>32-50 years</td>
<td>37-65 years</td>
<td>31-50 years</td>
<td>27-47 years</td>
<td>CdLS &gt;CdC**, Autism**</td>
</tr>
<tr>
<td>Paternal age, range and mean (SD)</td>
<td>32-48 years</td>
<td>30-65 years</td>
<td>31-48 years</td>
<td>28-47 years</td>
<td>--</td>
</tr>
<tr>
<td>% of primary caregivers married or living with a partner</td>
<td>87.6%</td>
<td>93.6%</td>
<td>66.6%</td>
<td>70%</td>
<td>--</td>
</tr>
<tr>
<td>% of families earning below £25,000</td>
<td>28.6%</td>
<td>33.3%</td>
<td>47.1%</td>
<td>75%</td>
<td>--</td>
</tr>
</tbody>
</table>

* p<.05  
** p<.01
Table 2. The number of mothers and fathers at or above clinical cut off levels for anxiety and depression.

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Number of mothers/females reaching clinical cut off</th>
<th>Number of fathers/males reaching clinical cut off</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Anxiety</td>
<td>Depression</td>
</tr>
<tr>
<td>Angelman Syndrome</td>
<td>10/14 (71.4%)</td>
<td>3/14 (21.4%)</td>
</tr>
<tr>
<td>Cri du Chat Syndrome</td>
<td>7/18 (38.9%)</td>
<td>4/18 (16.7%)</td>
</tr>
<tr>
<td>Cornelia de Lange Syndrome</td>
<td>5/15 (33.3%)</td>
<td>5/15 (33.3%)</td>
</tr>
<tr>
<td>Autism and ID</td>
<td>11/20 (55%)</td>
<td>3/20 (15%)</td>
</tr>
<tr>
<td>Normative population*</td>
<td>12%</td>
<td>4%</td>
</tr>
</tbody>
</table>

*Normative scores based on Crawford et al. (2001).
Table 3. Maternal and paternal outcomes (means and standard deviations) between the four etiology groups

<table>
<thead>
<tr>
<th>Measure</th>
<th>Maternal outcomes</th>
<th>Paternal outcomes</th>
<th>Post-hoc</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Angelman syndrome</td>
<td>Cornelia de Lange syndrome</td>
<td>Cri du Chat syndrome</td>
</tr>
<tr>
<td>HADS anxiety</td>
<td>11.71 (3.97)</td>
<td>8.93 (4.73)</td>
<td>9.49 (2.90)</td>
</tr>
<tr>
<td>HADS depression</td>
<td>8.57 (3.08)</td>
<td>7.30 (5.03)</td>
<td>7.36 (3.42)</td>
</tr>
<tr>
<td>QRS-F Family problems</td>
<td>10.61 (2.18)</td>
<td>5.86 (3.38)</td>
<td>7.43 (2.91)</td>
</tr>
<tr>
<td>GSSS</td>
<td>26.31 (8.17)</td>
<td>19.76 (8.78)</td>
<td>20.94 (7.23)</td>
</tr>
<tr>
<td>Positive Affect Scale</td>
<td>18.64 (6.89)</td>
<td>21.53 (10.84)</td>
<td>19.05 (9.41)</td>
</tr>
<tr>
<td>Positive Gain scale</td>
<td>6.93 (3.50)</td>
<td>5.67 (5.49)</td>
<td>7.39 (2.55)</td>
</tr>
</tbody>
</table>

* p<.05  
** p<.01  
†this difference was no longer significant once non-parametric tests were run.