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Mental health and well-being in mothers of children with rare geneitc syndromes showing chronic challenging behavior : A cross-sectional and longitudinal study

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Abstract

It is well documented that mothers of children with challenging behavior (CB) experience elevated levels of stress and that this persists over time, but less is known about the experience of mothers of children with rare genetic syndromes. This paper describes two studies, one cross-sectional and one longitudinal, comparing well-being in mothers of children with Angelman, Cornelia de Lange and Cri du Chat syndrome who have either shown chronic CB (*n*=18) or low/no CB (*n*=26) in the preceding seven years. The presence of chronic, long-term CB increased maternal stress but not depression or anxiety, and did not influence positive well-being. Stress relating specifically to their child's genetic syndrome reduced with age, highlighting the need for further exploration in this area.

Keywords: intellectual disabilities, syndrome, challenging behavior, mental health

Mental health and well-being in mothers of children with rare genetic syndromes showing chronic challenging behavior: A cross-sectional and longitudinal study

Introduction

Research focusing upon the mental health of mothers of individuals with intellectual disabilities (ID) has consistently reported elevated levels of maternal stress, depression and anxiety when compared to mothers of children without ID (Feldman et al., 2007; White & Hastings, 2004). Exploration of child factors has revealed that these elevated levels of maternal distress are not related to the severity of ID per se but to the behaviors displayed by the individual with ID (Bourke-Taylor, Pallant, Law, & Howie, 2012).

The knowledge base pertaining to behaviors that impact on stress and well-being of parents is largely derived from studies involving individuals with unknown etiology of ID or Autism Spectrum Disorder (see review by Hayes & Watson, 2013) using cross-sectional methodologies. Research examining the relationship between child behavior and parent mental health over time is more limited (Gray et al., 2011) with the few available studies documenting conflicting reports of improvement, stability or decline (Glidden & Schoolcraft, 2003; Lounds, Seltzer, Greenberg, & Shattuck, 2007; Esbensen, Rojahn, Aman, & Ruedrich, 2003). The complexity of the relationship between child behavior and parental mental health is highlighted by Woodman, Mawdsley and Hauser-Cram (2015) who suggest a longitudinal bidirectional relationship between parental stress and child behavior but with differing cross-over effect at different ages. In noting the range of limitations of the available research, which includes retrospective collection of parental mental health data (Flaherty & Glidden, 2000) or no consideration of child behavioral problems (Seltzer & Greenberg, 2001), Gray et

al. conclude that further longitudinal research is needed in order to more fully understand the relationship between behavioral problems and parental mental health.

With advances in genetic testing identifying an increasing number of genetic etiologies of intellectual disabilities, there has been increased interest in studying the behavioral phenotypes of syndromes, their molecular subgroups and their possible effects on family members (Miodrag & Peters, 2015). The majority of the work describing family stress in genetic syndromes to date relates to Down syndrome, although there has been recent increased interest in rarer syndromes (Cianfaglione et al., 2015; Close, Sadler, & Grey, 2015). To some extent, the experiences of mothers of children with rare genetic syndromes overlap with the experiences of all mothers of children with intellectual disabilities (Shearn & Todd, 1997; Udwin, Howlin, Davies & Mannion, 1998). However, there are a number of factors that are likely to be unique to mothers of children with rare genetic syndromes, many of which may be sustained and pervasive and lead to increased stress or anxiety. Using qualitative interviews, Griffith et al., (2011a) suggested that the lack of information about a syndrome, for both parents and medical professionals, leaves mothers feeling "in the dark" about the future and results in professionals being unaware of syndrome associated difficulties. This form of chronic stressor is not typically shared by mothers of offspring with more common causes of intellectual disability (Griffith et al., 2011a), highlighting the need for further research into the experiences of parents with rare genetic syndromes associated with intellectual disabilities. Given that the majority of the research into the mental health of mothers of children with rare genetic syndromes has focused on children aged 18 and younger, it is important that researchers begin to include adult children within their research.

In a large cross-sectional study, Adams et al. (2014) explored parental stress in a sample of mothers of children with nine rare genetic syndromes and compare these to mothers of children with autism. They note different patterns of between-syndrome differences for general stress and that relating to the genetic syndrome. Whilst this study clearly benchmarked the presence of elevated stress in mothers of children with a range of rare genetic syndromes, it was purely descriptive, only cross-sectional (and therefore can only describe the association with child age, but cannot identify if this is due to factors such as cohort effects) and did not investigate the influence of factors such as challenging behavior, which is particularly important given the high prevalence of challenging behaviors within some of the genetic syndromes.

In recognizing the need for further research into the mental health of parents of children aged 2–18 years with rare genetic syndromes, Griffith et al. (2011b) report on both paternal and maternal mental health within three syndromes, Angelman syndrome (AS), Cornelia de Lange syndrome (CdLS) and Cri du Chat syndrome (CdC). These three rare syndromes were selected because they share behavioral features, including severe ID and elevated rates of challenging behavior (Griffith et al., 2011b). By using standardized measures of anxiety, depression and stress, and by recruiting a well-documented high psychological distress comparison group (autism and ID), Griffith et al. were able to assess the relative degree of psychological distress in parents of children with rare syndromes who were showing clinically significant levels of challenging behavior. The results showed that mothers of children with all three syndromes, and fathers of children with AS and CdC syndromes, were more likely to report clinical levels of anxiety and depression than parents of children without ID. They also note that both mothers and fathers of children with AS had higher levels of stress than those parents of children with autism, CdC and CdLS. There were

no etiology-based differences on the measures of positive mental health. However, as their sample only contained individuals showing clinically significant challenging behavior, it is unknown whether it is the additional stress of parenting a child with a rare genetic syndrome, the high levels of challenging behavior, or a combination of the two, that is contributing to these elevated levels of parental psychological distress.

This paper consists of two studies which aim to answer the following research questions

- Study 1: Does the presence of challenging behavior affect mental health and well-being in mothers of children with rare genetic syndromes? This question will be answered using cross-sectional data, comparing measures of mental health and well-being in mothers of children who do show clinically significant levels of challenging behavior, with those that do not. The children have one of three genetic syndromes; AS, CdLS or CdC.
- Study 2: Do the elevated levels of psychological distress reported in mothers of children with rare genetic syndromes who are showing chronic challenging behaviour remain stable over time? Here, longitudinal follow-up data on maternal mental health from participants in the study by Griffith et al. (2011) are reported.

Based upon the previous literature on children with intellectual disabilities not associated with a rare genetic syndrome, it is hypothesized that there will be a positive relationship between the presence of challenging behaviour and poorer maternal mental health (elevated maternal anxiety and depression, elevated negative affect and/or reduced positive affect) which will remain present longitudinally.

By using both longitudinal and cross-sectional analyses, we can begin to address the complex relationship between maternal health, caring for a child with a rare genetic syndrome and chronic challenging behavior. Given the shared descriptions of the participants and the method across both studies, these are described in detail below before the results of each of the two studies are reported individually.

Method

Participants

Following ethical approval from the Coventry and Warwick NRES Committee and relevant R&D departments, participants were invited to take part via email or post. Participants completed the survey online or using pen and paper. Interview measures were undertaken over the telephone within two weeks of the questionnaires being completed. All parents were the biological parents of their children, with the exception of two who were long-term foster carers and one who was an adoptive parent. The term children is used throughout this paper to describe "offspring" but it is acknowledged that many of these children were adults at Time 2 of this study All of the participants' children spent at least part of the week living at home. Of the seven people who not living at home with their parents full-time, four lived in a group residential home and three lived in supported accommodation. Five of the seven people who spent part of their week away from home were in the Low/No group and one was from the Chronic CB group. All were noted to come home for weekends and some for "holidays". Two groups of participants are described in this paper based upon the presence or absence of challenging behavior in the six to seven years prior to data collection (see Figure 1); the Chronic CB and the Low/No CB group, each of which will be described below.

++Insert figure 1 about here++

The Chronic CB group

These participants were mothers originally recruited by Griffith et al. (2011), which was part of a larger study (Moss et al., 2013) focusing on the functioning of children with three rare genetic syndromes with clinically significant challenging behavior. At the time of original recruitment into Moss et al., all children: (1) had a diagnosis of either AS, CdLS or CdC; (2) were between 2 and 19 years of age; and (3) displayed self-injurious or aggressive behavior on at least a daily basis.

Of the 47 mothers described in Griffith et al. (2011), four had requested no further contact and four had moved out of England or Wales (the area covered by ethical approval). Three of the children (all from the CdLS group) had died. Therefore, 36 mothers from the original sample were invited to take part, of which 18 (50%) agreed to do so; eight had children with AS, four with CdLS and six with CdC.

The Low/No CB group

These mothers were recruited from a database of participants who had previously been involved in research. Participants were selected if they were mothers of children with one of the same three rare genetic syndromes as the Chronic CB group (AS, CdLS, CDC). However, the children in this group were not reported to have shown any clinically significant challenging behavior in a parallel study undertaken at approximately the same time as Griffith et al. (2011) data collection (in years 2007/8). Therefore, at the time of recruitment into this cross-sectional study (in 2014/15), all participants in the comparison group: (1) had a diagnosis of AS, CdLS or CdC and (2) were reported to show no self-injurious or aggressive behavior, or show these behaviors less than twice per week on a questionnaire administered between 2007 and 2008. Using the above criteria, 96 participants

(37 AS, 36 CdLS and 23 CdC) were invited to take part as the comparison group, of which 26 (27%) completed the questionnaires and interviews. Ten mothers of children with AS, ten with CdLS, and six with CdC participated.

Validation of participant grouping

Information gathered through the Challenging Behavior Interview (Oliver et al., 2003) confirmed that at time of the data collection (in 2014/15, hereafter referred to as Time 2), none of the participants in the Low/No CB showed clinically significant self-injurious or aggressive behavior (defined as a frequency of more than twice per week). This means that the Low/No CB group were not showing clinically significant CB in 2007/8 (hereafter referred to as Time 1) or at the Time 2 cross-sectional data collection (2014/15). In contrast, the Chronic CB group showed clinically significant CB at Time 1 and were reported to show clinically significant challenging behavior on the Challenging Behavior Interview at Time 2. At Time 1, 15 of the 18 Chronic CB participants showed self-injurious behavior of a frequency of at least once per hour. Of these 15, at Time 2, 12 were still reported to be showing self-injurious behavior at least hourly and three were showing the behavior at a frequency of at least once per day. At Time 1, 16 of the participants were reported to show aggression, of which eight were reported to show this behavior at least hourly, six at least daily and two at least once per week. Of these, the frequency remained stable for all but one participant who changed from showing the behavior weekly to daily.

Measures

The same measures were used at Time 1 and Time 2, focusing upon maternal well-being, challenging behavior and adaptive behavior. A demographic questionnaire was also used to collect information about children's and parents' ages, genetic or clinical diagnoses, gender, speech and mobility and living arrangements.

The Positive Gain Scale (PGS: Pit-ten Cate, 2003) is comprised of seven items which assess the direct positive aspects of having a child with a disability, such as "since having this child I feel I have grown as a person". Each item is rated on a 5-point Likert scale. These scales were reverse coded for this study, so the higher the score, the more positive gains reported by the participant. Internal consistency is good, with Cronbach's alpha being reported as .71 at Time 1 (Griffith et al., 2011) and .89 at Time 2. Pit-ten Cate (2003) notes good face and content validity.

The Positive Affect Scale (PAS) was derived by extracting the ten positive affect items from the Positive and Negative Affect Scale (PANAS; Watson, Clark, & Tellegan, 1988). Participants were presented with five descriptive words, such as "strong" and "interested" and were asked to rate the extent to which they felt this way over the past week on a Likert-type scale. A higher score indicates more positive affect. <remove for blind review> report Cronbach's alpha to be .91 for mothers of children with rare genetic syndromes and it is .92 at Time 2. Watson et al. (1988) reported a test-rest reliability of .68 over a period of eight weeks.

The Parent and Family Problems Subscale from the Questionnaire on Resources and Stress – Short Form (QRSF; Friedrich, Greenburg, & Crnic, 1983), was used to measure general stress associated with raising a child with intellectual disabilities. As Griffith et al. (2011), the five items assessing depression were excluded to reduce potential overlap. Mothers were asked to circle "true" or "false" on 15 items, such as "other members of the family have to do without things because of N". The Kuder-Richardson coefficient for this version of the questionnaire was .85 at Time 2. Reliability and convergent validity for the full subscale are described as good for parents of children with autism (Hastings & Brown, 2002; Honey, Hastings, & McConachie, 2005), but no studies have explored the reliability or validity (other than Kuder-Richardson coefficients) for the subscale with the depression items removed.

The Hospital Anxiety and Depression Scale (HADS; Zigmond & Snaith, 1983) was used to assess parental mental health. Each item is scored on a four point Likert scale from 0–3, resulting in a maximum possible score of 21 for both the depression and anxiety subscale. This was chosen b Griffith et al. (2011) to allow for direct comparison to a sample of mothers of children with autism. It has good test-retest reliability and concurrent validity is reported at subscale and total score level (.49–.83), including for parents of children with neurodevelopmental disabilities (Bjelland, Dahl, Haug, & Neckelmann, 2002). A score of eight or above is considered to represent "clinical" levels of depression or anxiety. In the present sample at Time 2, Cronbach's alpha was strong (.91 for both subscales).

The Genetic Syndrome Stressors Scale (GSSS: Griffith et al., 2011a) was used to measure parental stressors relating to rare genetic disorders. Parents were asked to read statements, such as "not having access to professionals who have knowledge about my

child's condition", and asked to rate how stressful they have found this condition in the last six months. In the present sample (Time 2), Cronbach's alpha was .88. Griffith et al. (2010a) report good face validity and moderately good concurrent validity.

The Vineland Adaptive Behavior Scales 2nd Edition (VABS-II: Sparrow, Cichetti, & Balla, 2005) is a semi-structured interview used to assess three domains: Socialization, Daily living skills and Communication. The Motor Skills or Maladaptive Behavior Domains were not administered. Mothers were interviewed over the telephone within two weeks of completing the questionnaire. The VABS-II manual reports good test-retest reliability, with correlations ranging from .80-.95 and inter-rate reliability correlations ranging from .75–.85.

The Challenging Behavior Interview (CBI: Oliver et al., 2003) assesses the presence and severity of specified forms of behavior in people with intellectual disability. Respondents first identify topographies of behavior displayed in the last month, before answering 14 questions on the characteristics of the behavior, such as frequency, duration, and necessary response. The authors report good inter-rater, test-retest reliability and content validity. This was used to document the level of challenging behavior and therefore validate membership in either the Chronic CB or Low/No CB group. Oliver et al. report good mean kappa indices for inter-rater reliability (.67) and moderate for test–retest reliability (.86).

Study 1: Exploring the impact of challenging behavior on mental health in mothers of children with rare genetic syndromes

This study aims to explore whether the presence of challenging behavior is associated with maternal mental health in mothers of children with AS, CdLS and CdC in order to answer the first research question; does the presence of challenging behavior affect mental health and well-being in mothers of children with rare genetic syndromes? This question will be explored using cross-sectional data, comparing maternal mental health in mothers of children with a rare genetic syndrome who have or have not shown clinically significant levels of challenging behavior in the five to seven years preceding data collection.

Participants

Table 1 shows the demographic information of the sample at time of the cross-sectional data collection (Time 2, 2014-2015), including the ages and gender distribution of the children and the ages of their mothers. All participant's children had their rare genetic syndrome either made or confirmed by a clinical geneticist, pediatrician or neurologist.

Table 1*: Means, SDs, Ranges and Statistical Comparisons of Demographic Characteristic by Group*

	Chronic CB *n* = 18		Low/No CB *n* = 26		Group differences	
	M	*SD*	*M*	*SD*	t	p
Child age in years	16.2	4.1	23.0	8.0	3.4	.002
Vineland Daily Living Skills Standard Score	34.5	9.8	29.5	11.7	-1.5	.16
Maternal age in years	48.2	6.8	51.5	10.9	1.1	.27
	n	%	*n*	%	χ^2	p
Males	10	55.6	12	46.2	0.24	.43
Speech (verbal)	9	50.0	15	57.7	0.25	.76
Mobility (mobile)	15	83.3	18	69.0	1.13	.48
Accommodation (Living at home full-time)	17	94.4	65	65.0	3.80	.60

A series of one-way t-tests and chi-square tests were conducted on the demographic variables across the two groups. The Low/No CB group was significantly older than the Chronic CB group, so in order to control for this, any cross-sectional comparative analyses were repeated with the age of the child entered as a covariate.

Results

In order to explore the impact of chronic challenging behavior on maternal anxiety, depression, stress, positive affect, positive gain and reported stress relating to the genetic syndrome, a MANOVA was conducted with group (Chronic CB, Low/No CB) as the independent variable and the measures of maternal mental health (anxiety, depression, stress, GSSS score, positive affect and positive gain) as the dependent variables. There was a statistically significant difference in maternal mental health between the Chronic CB and

Low/No CB group, (F(6,35) = 2.77, p = .03). The data and between-subjects comparisons are reported in Table 2.

Table 2. *Maternal Mental Health (means and standard deviations) in the Chronic and Low/No CB Groups.*

	Chronic CB Group (n = 18)		Low/No CB Group (n = 25)		MANOVA Post-hoc Statistic *df* (1,40)		Effect Size	MANCOVA Post-hoc Statistic *df* (1,39)	
	M	*SD*	*M*	*SD*	F	*p*	d	F	*p*
HADS Anxiety	9.73	4.62	8.56	5.16	0.46	.50	0.24	0.28	.60
HADS Depression	7.28	3.49	6.12	5.04	0.90	.35	0.26	0.53	.47
QRS-F Family Problems	10.05	2.90	6.20	3.73	12.4	.001	1.13	7.02	.009
GSSS	23.72	7.92	15.72	8.60	8.88	.005	0.96	3.61	.07
Positive Affect	31.83	8.10	32.01	9.38	0.004	.95	0.02	0.06	.81
Positive Gain	13.83	9.19	12.82	7.80	0.06	.81	0.12	0.06	.81

The results show that mothers of children with AS, CdLS and CdC whose children show chronic challenging behavior experience elevated levels of general stress (QRSF-F) and report higher levels of stress relating to their child's rare genetic syndromes (GSSS) than mothers of children with the same genetic syndromes who are not showing challenging behavior. Both of these results have a large effect size. There were no between-group differences on levels of anxiety, depression, positive affect or positive gain. The scores on the HADS indicate that within the Low/No group, 50% had scores indicative of clinical levels of anxiety and 30.8% had scores indicative of clinical levels of depression. Within the Chronic CB group, 66.7% had scores indicative of clinical levels of anxiety and 38.9% had scores indicative of clinical levels of depression.

As the ages of the children differed between the two groups, the MANOVA was repeated with age of the child added as a covariate. The overall MANCOVA was not significant ($F(6,34) = 1.55$, $p = .19$). The between group differences on the measures of depression, anxiety, positive affect and positive gain all remained non-significant. With age as a covariate, the difference on the measure of maternal stress, QRSF, remained significant, but the difference on the measure focusing upon stress specifically relating to the rare genetic syndrome (GSSS) was not significant. Pearson's correlations show that the GSSS total score is significantly negatively correlated with child age ($r = -.44$, $p = .003$).

Study 2: Exploring the impact of long-term challenging behavior on mental well-being in mothers of children with rare genetic syndromes

Study One identified that child age influences the relationship between the presence of challenging behavior and maternal stress. However, the data were cross-sectional and would benefit from further exploration using longitudinal designs. This study will document the longitudinal progression of mental health in 18 mothers of children with a rare genetic syndrome showing clinically significant levels of challenging behavior by comparing the data collected for the *Chronic CB* group in this study (Time 2) to that documented at Time 1 (Griffith et al., 2011)).

Participants

The average time between data collection at Time 1 and Time 2 was 6.7 years (range 5.7-7.7 years). Demographic details of Time 1 and Time 2 are documented in Table 3 below.

Table 3 Means, SDs, Ranges of Demographic Characteristics at Participants in Chronic CB Group (n=18) at Time 1 and Time 2

	Time 1		Time 2	
	M	*SD*	*M*	*SD*
Child age	9.5	4.2	16.2	4.1
Vineland Daily Living Skills Standard Score	42.1	10.3	34.5	9.8
Carer age	41.8	6.9	48.2	6.8
	n	%	*n*	%
Speech (verbal)	9	50.0	9	50.0
Mobility (mobile)	15	83.3	15	83.3
Accommodation (Living at home full-time)	18	100	17	94.4[1]

[1] One participant had moved into part-time supported accommodation and returned to the family home at weekends.

Results

Comparisons were made between those who had responded to the invitation to take part in Time 2 and those who had not responded. There were no significant differences between those who did and did not respond on child ($t(34) = -0.46$, $p = .65$) or maternal age ($t(34) = -0.74$, $p = .47$), scores on the Vineland Communication ($t(34) = 0.43$, $p = .67$), socialization ($t(34) = 1.80$, $p = .08$) or Daily Living Skills ($t(34) = 1.96$, $p = .06$) domains nor on measures of maternal anxiety ($t(34) = 0.84$, $p = .41$), depression, ($t(34) = 1.56$, $p = .13$), stress ($t(34) = -1.80$, $p = .08$), positive gain ($t(34) = 0.53$, $p = .60$) or positive affect ($t(34) = -0.43$, $p = .67$).

In order to explore levels of anxiety, depression, stress and positive mental health over time in parents of individuals with *Chronic CB*, repeated measures ANOVAs were conducted. As time between the two data collection points varied between participants, the analyses were repeated with the time between the two data collection points entered as a covariate. The results are presented in Table 4.

Table 4. *Maternal Outcomes (means and standard deviations) at Time 1 and Time 2 for the Chronic CB Group*

	Time 1		Time 2		ANOVA *df* (1,17)			ANCOVA *df* (1,16)	
	M	*SD*	*M*	*SD*	F	*p*	Effect Size d	F	*p*
HADS Anxiety	9.83	3.47	9.72	4.62	0.30	.87	0.26	1.60	.23
HADS Depression	7.19	2.90	7.28	3.49	0.01	.91	0.05	0.77	.39
QRS-F Stress	9.38	2.72	10.10	2.90	1.24	.28	0.52	2.57	.13
GSSS	23.40	7.08	23.70	7.92	0.05	.82	0.11	0.00	.99
Positive Affect Scale	27.99	7.81	31.80	8.10	3.84	.07	0.92	1.42	.25
Positive Gain Scale	13.05	5.80	13.80	9.20	0.15	.71	0.18	0.21	.65

Table 4 highlights that there was no statistically-significant change in any of the measures over time. However, the effect size calculation shows a small effect size for the change in anxiety scores, a medium effect size for the increase in stress scores and a large effect size for the increase in the positive affect scale.

These results show that the elevated levels of maternal anxiety, depression and stress reported in mothers of individuals with rare genetic syndromes who have *Chronic CB*

continue to be present six to seven years after they were initially reported. The results remain consistent even when time between the two data collection points is entered as a covariate.

At Time 1, 13 mothers (72.2%) had scores indicative of clinical levels of anxiety and 8 (44.4%) had scores indicative of clinical levels of depression. At Time 2 (as documented above), 12 (66.7%) had scores indicative of clinical levels of anxiety and 7 (38.9%) had scores indicative of clinical levels of depression.

Discussion

This paper combined cross-sectional and longitudinal data to explore whether elevated levels of stress reported in parents of children with rare genetic syndromes are associated with any additional stress of parenting a child with a rare genetic syndrome, the high prevalence of challenging behavior, or a combination of the two.

The two studies presented in this paper revealed three main findings: (1) The presence of chronic, long-term challenging behavior is associated with increased levels of maternal stress but not increased levels of depression or anxiety in mothers of children with AS, CdC, and CdLS; (2) the association between chronic challenging behavior and elevated levels of stress specifically relating to the child's genetic syndrome is influenced by the child's age, with GSSS total score reducing as child age increases; and (3) the elevated mental health difficulties associated with parenting a child with a rare genetic syndrome and chronic, clinically significant challenging behavior remain present even after six to seven years. This provides a clear example of where a non-significant result (i.e., no change in levels of mental

health measures) provides clinically important information. It is also important to acknowledge the non-significant but large effect size result of an increase in positive affect between Time 1 and Time 2.

Although somewhat unsurprising given the well-documented chronicity of challenging behavior, it is important to highlight that all of the parents from Griffith et al. (2011) study reported that their child continued to show clinically significant challenging behavior five to seven years after their initial assessment. Although there are well-established behavioral treatments, the literature suggests that parents find them complex and prone to attrition (Murphy, Oliver, & Corbett, 1993; Oliver, Murphy, & Corbett, 1987), limiting their clinical efficacy. Given the strong association between challenging behavior, health problems, reduced quality of life and limited access to social activities, the finding that challenging behavior remained present is concerning.

The results of the MANCOVA highlight the influence of child age on the relationship between challenging behavior and maternal stress relating specifically to stress associated with a child's genetic syndrome. Although correlations suggest that the GSSS score decreases (reflecting lower stress) as the child ages, it is important to interpret these data alongside those of the longitudinal study, where the GSSS scores did not differ between Time 1 and Time 2. This may suggest that either change in GSSS scores occurs over a period longer than the average 6.7 years between these two data collection points, or that the result is an artifact of the cross-sectional design used. If the change in scores occurs over a longer period of time (between childhood and adulthood), this may reflect the fact that the questionnaire was derived with a strong child focus. Potentially, this means that the issues causing stress when parenting an adult with a rare genetic syndrome may differ from those present when the child

is younger, and consequently these are not addressed in this questionnaire. Behavioral presentations and phenotypes of AS (Adams, Horsler, Mount, & Oliver,, 2015) and CdLS (Cochran, Moss, Nelson, & Oliver, 2015), change from childhood into adolescence and adulthood highlighting the need for more adult-based research in rare genetic syndromes, ideally studies using longitudinal methodologies to reduce the risk of potential confounds from cross-sectional designs.

These results need to be considered alongside a number of methodological limitations. Most notably, as is common in studies recruiting rare syndromes, the sample size was relatively small (especially in the Chronic CB group), limiting the extent to which any between-syndrome group comparisons could be completed. Given that Griffith et al (2011b) found that mothers of children with AS were significantly more stressed than mothers of children with autism, CdLS or CdC, a larger sample would have allowed for exploration as to whether such differences remain in a longitudinal design. With the sample size available, there are also a large number of maternal mental health measures. Although robust group differences were noted in the cross-sectional study, the small sample size may have limited the power to reveal any further group differences. The power calculations highlight the strength of the differences in the cross-sectional comparison design, with Cohen's d reflecting a large effect size. The power calculations also suggest that the small sample size is resulting in a Type II error for the stress and positive affect scales. This highlights the importance of denoting effect sizes with rare syndrome research, as by the very nature of their rarity, it is difficult to collate large sample sizes, especially in longitudinal designs. Given the large effect size for the Positive Affect Scale in the longitudinal scale (0.92), further research and focus is needed to explore positive mental health in mothers of children with rare genetic syndromes. This is important for potential parenting interventions, as

parents who have more positive mental well-being when their child is young experience less stress later in life (Paczkowski & Baker, 2008). Future research should therefore not only consider how maternal positive affect may change with children's ages, but also how this can be enhanced and how it relates to other measures of maternal well-being (see Horsley & Oliver, 2015 for a discussion of positive mental health in this population).

Due to having only two data points, it is not possible to document what happened to behavior and parental stress in between the two assessment times. It may be that there were substantial periods of time where the challenging behavior in the Chronic CB group significantly reduced, or significantly increased in the Low/No CB group, or times when maternal mental health was significantly different from that reported in these studies. Finally, it is recognized that the majority of mothers in this study were members of a national syndrome support group (which may result in a recruitment bias), it was unknown if the mothers were the primary caregiver, and there was a lack of confirmatory genetic data.

Although the association between challenging behavior and maternal mental health difficulties is also well documented as persisting over time (Dyson, 1993), to our knowledge, this is the first paper to document this longitudinally in these three syndromes, and to use both longitudinal and cross-sectional designs to control for the presence of challenging behavior within the context of a rare genetic syndrome. It is also the first (to our knowledge) which, over time, quantitatively measures positive well-being and perceptions of positive gain in parents of children with rare genetic syndrome and intellectual disabilities. The positive outcomes did not differ between the groups and remained stable over time in the CB group, supporting the notion that positive outcomes may be relatively independent of child

characteristics (Hastings & Taunt, 2002). Finally, this is the first paper to report longitudinal data on the measure of stress relating to genetic syndromes (GSSS).

Future work should begin to explore the interaction between parental mental well-being and challenging behavior within families of children with rare genetic syndromes, exploring models such as that purported by Woodman et al. (2015). There is also a need for research which includes or focuses upon adults with rare genetic syndromes and how behavior, behavioral phenotypes, family adaptation and parental well-being may change over time. Ideally such work would be longitudinal, having better control over individual differences and environmental factors. Finally, as this paper highlights the chronicity of challenging behavior and its long-standing association with parental well-being, more research is needed to explore services and interventions accessed, the barriers or motivators to engage in such interventions and the perceived level of effectiveness at helping to reduce challenging behaviors.

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