Genetic syndromes in the family : child characteristics and parenting stress in Angelman, CHARGE, Cornelia de Lange, Prader-Willi, and Rett syndrome

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3 Parenting stress in mothers with a child with Rett syndrome

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ABSTRACT

Parenting stress can have severe negative consequences. This study investigates maternal parenting stress in families with a child with Rett syndrome (RS) and the relationship with child characteristics. Twenty-four mothers of a child (2-17 years) with RS participated. Four questionnaires were used: the Nijmegen Parenting Stress Index-Short, Vineland Screener 0-6, Developmental Behaviour Checklist, and Rett Syndrome Behaviour Questionnaire. Parenting stress was high in 46% of the mothers. Maternal parenting stress was not related to the child's age, adaptive functioning, and presence/absence of the autistic disorder. General behavioural problems correlated positively with parenting stress. Of RS specific behaviours, only general mood problems correlated positively with parenting stress. Having a child with RS is a risk factor for high maternal parenting stress. Especially when children show behavioural problems, support is needed. Future studies should focus on processes that lead to different outcomes in these families, as not all mothers perceive much stress.

INTRODUCTION

Rett syndrome (RS) is a neurodevelopmental disorder whereby *MECP2* gene mutations exist in most persons with the syndrome (Percy, 2008). RS is almost exclusively present in females. A classical variant and certain atypical ones are distinguished. One of the hallmarks of the syndrome is a seemingly normal early development, followed by the loss of acquired developmental skills usually at the age of one to two years. After this regression period, some improvement in later years does occur (Hagberg, 2002). Most females with RS function on the severe to profound level of intellectual disability (Demeter, 2000).

The characteristics of the syndrome make the occurrence of stress in family life imaginable. High levels of parenting stress are an important target for intervention; such distress can lead to withdrawn parenting and distressed parents are less likely to promote the child's development optimally (Deater-Deckard, 2004). It can have negative consequences for parents as well, such as poorer physical health (Oelofsen & Richardson, 2006) and depression (Singer, 2006). In some parents with a child with RS high stress levels were found, although most parents reported stress in the normal range. No relation

between parenting stress and the child's age, adaptive level or cognitive functioning was found (Perry, Sarlo-McGarvey, & Factor, 1992). In contrast, psychosocial stress in mothers was higher when more RS specific behaviour was present, in particular general RS mood problems, behaviours indicative of fear/anxiety, RS specific night-time problems, and total amount of typical RS behaviour (Sarimski, 2003).

The aim of the present study is to expand the knowledge on parenting stress in mothers with a child with RS and to investigate which child characteristics are related to maternal parenting stress. This knowledge may contribute to more specific support for these families. As far as we know, this is the first study to relate maternal parenting stress to general behavioural problems of children with RS. The relation between maternal parenting stress and a co-morbid autistic disorder in RS will also be explored. The combination of a child with ID and autism is more distressing than having a child with ID only (Blacher & McIntyre, 2006). However, it is unclear whether this also applies to mothers with a child with RS.

METHOD

Participants

Participants were 24 families with a daughter with RS. They were part of a larger study on RS. In this paper only children between the age of 2 to 18 years were included for whom the mother filled out the questionnaire on parenting stress to obtain a relatively homogeneous group. They make up 46% of the larger sample, see Wulffaert, Van Berckelaer-Onnes, and Scholte (2009) for more details on the procedure and participants. Mean age was 9.2 years (*SD* = 4.74; range 2.4 – 17.2). The classical variant was present in 18 females of whom 16 had a *MECP2* mutation, 1 did not have a *MECP2* mutation and in 1 female no genetic testing was carried out. In five participants the atypical RS variant was present, all had a *MECP2* mutation. For one female the RS variant was unknown, but a *MECP2* mutation was confirmed.

Research Instruments

The *Nijmegen Parenting Stress Index-Short* (NPSI-S; De Brock, Vermulst, Gerris, & Abidin, 1992) is an official translation and adaptation of the Parenting Stress Index by Abidin (1983, as cited in De Brock et al., 1992). It measures parenting stress in families

with children from approximately 2 to 13 years. Since the level of functioning of the RS females not exceeded this level, the instrument was considered appropriate for the purpose. A total score is computed and classified into seven norm categories, for mothers and fathers separately, defining parenting stress level. Dutch non-clinical and clinical norms are available; the non-clinical norm group, based on families of the normal population, was used. Psychometric properties are reasonable to good (De Brock et al., 1992).

The *Vineland Screener 0-6 years* (VS 0-6; Scholte, Van Duijn, Dijxhoorn, Noens, & Van Berckelaer-Onnes, 2008) is a Dutch screening instrument adapted from the Vineland Screener by Sparrow, Carter, and Cicchetti (1993). The VS 0-6 measures the level of adaptive functioning of children up to the age of six or older people with comparable levels of functioning. An adaptive behaviour composite score is based on the domains communication, daily living skills, socialisation, and motor skills. The instrument has good reliability and validity (Scholte et al., 2008).

The Dutch version (Koot & Dekker, 2001) of the *Developmental Behaviour Checklist-Primary Carer* (DBC-P; Einfeld & Tonge, 2002) assesses emotional and behavioural problems in children with intellectual disabilities. A total behaviour problem score is computed together with five subscale scores (disruptive/antisocial behaviour, self-absorbed behaviour, communication disturbance, anxiety, social relating problems). Psychometric properties are satisfactory to good (Koot & Dekker, 2001). The DBC-P has an additional autism screening algorithm which reliably screens for the autistic disorder (Einfeld & Tonge, 2002).

The *Rett Syndrome Behaviour Questionnaire* (RSBQ; Mount, Charman, Hastings, Reilly, & Cass, 2002) describes behavioural and emotional features typical for RS. A Dutch translation was developed for this study. A total score is computed together with eight subscale scores (general mood, breathing problems, hand behaviours, repetitive face movements, body rocking and expressionless face, night time behaviours, fear/anxiety, walking/standing). Is has good psychometric properties (Mount et al., 2002).

All questionnaires have been processed conform the instructions of the official manuals and for the RSBQ conform the related article by Mount et al. (2002).

Data-analysis

The relationships between maternal parenting stress and child characteristics were determined by correlational analyses in SPSS 16.0. For the child characteristic 'presence of the autistic disorder' a comparative analysis of means was used. An alpha of .05 was chosen for all analyses. In case of non-normality following the Saphiro-Wilks test, non-parametric variants for *t*-tests and Pearson correlations were carried out, i.e. Mann-Whitney tests and Spearman correlations. Univariate outliers were given the next highest score plus or minus one, depending whether the outlier was at the higher or lower end. Effect sizes of $r = .10$ were seen as small, $r = .30$ as medium, and $r = .50$ as large (Field, 2009).

RESULTS

In Table 3.1 the perceived parenting stress in the participating mothers is compared to the non-clinical norm group. Overall, parenting stress was high in mothers with a child with RS. Although some mothers perceived stress levels categorised as very low to below the mean (5; 20%), nearly half of them (11; 46%) experienced high to very high stress.

Table 3.1 *Parenting stress in mothers with a child with Rett syndrome (n = 24)*

Maternal parenting stress NPSI-S norm category non-clinical norm group		**Mothers of a child with Rett syndrome % (*n*)**
Category	Percentiles in norm population	
Very low	0% - ≤ 5% (5%)[a]	4% (1)
Low	5% - ≤ 15% (10%)	4% (1)
Below the mean	15% - ≤ 35% (20%)	12% (3)
Mean	35% - ≤ 65% (30%)	17% (4)
Above the mean	65% - ≤ 85% (20%)	17% (4)
High	85% - ≤ 95% (10%)	**17% (4)**
Very high	95% - ≤ 100% (5%)	**29% (7)**

Note. NPSI-S = Nijmegen Parenting Stress Index-Short.
[a] Percentage of total population between brackets

The adaptive level of functioning was very homogeneous as expected, ranging from 3 to 14 months ($M = 7.9$ months, $SD = 3.19$). For both the DBC-P and RSBQ the number of items per subscale differ; to make mean scale scores comparable within the

instruments, scores were standardised with a possible range between 0 to 2 (see Table 3.2). DBC-P subscales self-absorbed behaviour and the autism screening algorithm received the highest mean scores. The least problems were mentioned on the disruptive/antisocial and communication disturbance subscales. On the RSBQ by far the highest score was measured on the hand behaviours scale, the lowest score on the night-time behaviour scale. According to the DBC-ASA 11 children did not need further screening for the autistic disorder, whereas in 13 children the autistic disorder was suspected to be present and further individual assessment was needed.

Table 3.2 *Standardised mean scores and correlations between raw maternal parenting stress scores and behavioural problems measured with the DBC-P and RSBQ (n = 24)*

	M	***SD***	***r***	***p***
DBC-P				
Self-absorbed	.63	.26	.59	<.01**
Autism screening algorithm	.61	.33	.53	<.01**
Social relating	.51	.31	.42	.04*
Total Problem Behaviour Score	.40	.21	.62	<.01**
Anxiety	.38	.35	.49	.02[a]*
Communication disturbance	.23	.23	.39	.06[a]
Disruptive/antisocial	.21	.20	.49	.02[a]*
RSBQ				
Hand behaviours	1 .51	.37	-.09	.67[a]
Fear/anxiety	.97	.46	.18	.39
RSBQ total	.90	.33	.19	.37
General mood	.87	.49	.48	.02*
Breathing problems	.83	.66	.04	.87[a]
Body rocking and expressionless	.82	.39	-.02	.91
Repetitive face movements	.75	.58	.04	.86[a]
Walking/standing	.63	.59	.09	.70[a]
Night-time behaviours	.47	.40	.14	.51[a]

Note. DBC-P = Developmental Behaviour Checklist; RSBQ = Rett Syndrome Behaviour Questionnaire.
[a] = Spearman correlation.
* $p < .05$, ** $p < .01$

Maternal parenting stress was not significantly nor substantially related to the child's age (r_s = -.19, p = .37), neither was there a significant relation between stress and

adaptive functioning (r = .18, p = .39). All DBC-P (sub)scales, except communication disturbances, related significantly with maternal parenting stress. These were positive correlations and ranged from medium (r^2 = .38) to small effects (r^2 = .18) (see Table 3.2). On the RSBQ only the general mood subscale related significantly and positive to maternal parenting stress with a small effect size (r^2 = .23). Maternal parenting stress was not related to the presence of autistic disorder according to the t-test for unequal variances (t (16) = -1.41, p = .18).

DISCUSSION

Raising a child with RS places mothers at risk for high levels of parenting stress; nearly half of them reported high to very high levels. However, there are also mothers who do not perceive heightened stress levels. The child's age and level of adaptive functioning does not influence the level of maternal parenting stress. These results are in line with the study by Perry et al. (1992). In the current study there appeared strong positive relationships with several specific behavioural problems, with medium sized effects for the total problem behaviour score, self-absorbed behaviour, and autism screening algorithm. Behaviours specific for RS were not related to parenting stress, except a positive relation with more general mood problems. Finally, although a significant correlation was found between maternal parenting stress and the autism screening algorithm, there was no difference for parents with a child scoring above versus below the cut off for autistic disorder. Thus, maternal parenting stress is comparable in children who presumably have a co-morbid autistic disorder versus those who do not.

The absent relationship between stress and adaptive abilities may be caused by the lack of variation in the children's level of functioning; in our sample nearly all had abilities below the developmental age of one year. In studies into other genetic syndromes, behavioural problems in general appeared the strongest predictor for parenting stress (Hodapp, 1999). The current study suggests that this pattern also exists in families with a child with RS. Overall RS specific behaviours were not related to maternal parenting stress. We hypothesize that these characteristic behaviours are nowadays so well known to belong to the syndrome, also for parents, that they might no longer induce much stress. Finally, although maternal parenting stress was related to behaviours indicative of the autistic disorder, there was no difference in stress between mothers with a child without

the autistic disorder and those who needed further individual assessment for it. Thus, for maternal parenting stress the *amount* of autistic behavioural problems seems more distressing. See Wulffaert, Van Berckelaer-Onnes, and Scholte (2009) for a further discussion of the controversial issue of a co-morbid autistic disorder in RS.

The relationship between RS specific behaviour and parental perceptions and well-being remains unclear. In the current study an association was found between RS general mood problems and maternal parenting stress. Other studies reported relationships between specific RS behaviour and more broadly defined psychological stress, and physical and mental health (Laurvick, Msall, et al., 2006; Sarimski, 2003). However, the results differed on which *specific* RS behaviours were relevant for parental perceptions. Future studies are needed to fully understand the impact these RS behaviours have on parents.

One of the limitations of our study is the small sample size which results in problems with statistical power. Also, the current study has a cross-sectional design. For persons with intellectual disabilities, results are mixed whether the child's behavioural problems cause parenting stress or whether there is a bi-directional effect (Hassall & Rose, 2005; Hastings & Beck, 2004; Olsson, 2008). Long-term follow-up studies are thus needed to investigate this pathway in RS. Furthermore, parental and environmental characteristics such as (in)formal support and parental coping strategies are characteristics influencing the outcomes of the stress process (Hassall & Rose, 2005; Perry, 2004). These factors should be incorporated in future studies to give a more coherent description of the families with RS. Finally, we follow Olsson's (2008) view that in future studies it is important to focus on the *processes* that lead to different outcomes in these families. Why do some families with a child with RS adapt well to their specific situation and others do not? There is still a lot to discover on causality and influencing risk and protective factors in research in families with RS.

The finding that parenting stress is high in nearly half of the mothers should raise awareness on the need for support for these families. When the child shows behavioural problems, parents should get additional support to manage them with reduced parenting stress as a consequence (Hastings & Beck, 2004). Laurvick, Msall, et al. (2006) found that in RS lower parental stress levels are associated with better mental health of mothers. Support for the child will thus benefit the health of parents as well. As there is no relation between stress and the child's age, support should be a continuous process and not only

limited to the early years, which can be so devastating in these families. Finally, we want to incorporate the advice by Sarimski (2003) and Laurvick, Msall, et al. (2006) that support in these families should also focus on the challenges caused by physical disabilities in RS (e.g. the feeding and dressing process) and underline the positive impact on the family system when mothers have time for own activities beside caretaking, such as having work outside the house or free time. The challenges these families face are many and deserve professional and specified support.