

## Parenting Stress in CHARGE Syndrome and the Relationship with Child Characteristics

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**Abstract** This study investigates the parental perception of stress related to the upbringing of children with CHARGE syndrome and its association with behavioral and physical child characteristics. Parents of 22 children completed the Nijmegen Parenting Stress Index-Short, Developmental Behavior Checklist, and Dutch Vineland Screener 0-12 and reported their child's problems with hearing, vision and ability to speak. Parenting stress was high in 59% of the subjects. Behavioral problems on the depression, autism, self-absorbed and disruptive behavior scales correlated positively with parenting stress. A non-significant trend was found, namely higher stress among the parents of non-speaking children. No associations were found with other child characteristics, i.e. level of adaptive functioning and intellectual disability, auditory and visual problems, deafblindness, gender, and age. Raising a child with CHARGE syndrome is stressful; professional support is therefore essential for this population. More research into other possible influencing characteristics is needed to improve family-oriented interventions. Since CHARGE is a rare syndrome, closer international collaboration is needed, not only to expand the group of study subjects to increase statistical power, but also to harmonize research designs and measurement methods to improve the validity, the reliability, and the generalization of the findings.

**Keywords** Behavioral phenotype · CHARGE syndrome · Parenting stress

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CHARGE syndrome is a genetic disorder in which multiple anomalies are present from birth. The acronym is derived from the combination of the following problems: **C**oloboma of the eyes, **H**ear defects, **A**tresia of the choanae, **R**etardation of growth and/or development and/or central nervous system anomalies, **G**enital hypoplasia, **E**ar anomalies and/or deafness (Pagon et al. 1981). At present, the criteria of Blake et al. (1998) and Verloes (2005) are usually used to diagnose the syndrome. These sets differ in some aspects, but both make use of rules about the number of ‘major’ and ‘minor’ signs needed for a CHARGE diagnosis. In addition to the clinical criteria, presence of a *CHD7* gene mutation on chromosome 8 is another way to establish the diagnosis (Vissers et al. 2004). According to a recent review, physical problems in many persons, besides those mentioned in the acronym, include vestibular problems, gastro-oesophageal reflux, facial paralysis, and feeding and swallowing problems (Sanlaville and Verloes 2007). For those suffering from the syndrome, impairments affect all senses and as a result have a severe impact on development (Brown 2005). Incidence has been estimated to range between 1:8,500 to 1:12,500 live births (Sanlaville and Verloes 2007).

Persons with CHARGE syndrome vary widely in the combination of physical problems present as well as their level of functioning and behavioral characteristics (Blake et al. 2005; Vervloed et al. 2006). The level of functioning ranges from profound intellectual disability (ID) to normal intelligence, but a substantial proportion seem to function in the lower range (Johansson et al. 2006; Salem-Hartshorne and Jacob 2005; Smith et al. 2005). Behavioral problems are often mentioned but the behavioral phenotype has not yet been completely defined. Self-injurious behavior, sleep problems, hyperactivity, irritability, attention problems, tactile defensiveness, adherence to routines, and stereotypical behaviors have been described (Blake et al. 2005; Graham et al. 2005; Johansson et al. 2006). Results regarding the occurrence of aggression are contradictory (Blake et al. 2005; Graham et al. 2005; Johansson et al. 2006). The behavioral problems seem to be more manifest in older persons (Hartshorne and Cypher 2004; Vervloed et al. 2006). Some studies, however, report low rates of behavioral problems (Graham et al. 2005; Smith et al. 2005).

Virtually all research has focused only on the child with CHARGE syndrome. Although this is inherent to the issue at stake, children develop in interaction with the environment and as such the parents play a vital role for these vulnerable children. Therefore, the way parents experience the childrearing situation needs to be considered. This may ultimately lead to better support for the family system. One way to describe the perception of parents is to measure level of parenting stress. Perry (2004) designed a model to depict the factors that influence the development of stress in families with a child with a developmental disability. It consists of four components, each divided into two domains. The first component in the stress process is the *stressor*, which can be divided into (1) child characteristics versus (2) other life stressors (e.g. divorce). Secondly, the *resources* of the family are divided into (1) family system resources (e.g. socio-economic status), and (2) personal resources of the parent (e.g. coping style). Thirdly, the *support* a family receives can be from (1) a professional service or (2) an informal system. The resources and

support systems act upon the influence of the stressor on parenting stress. The fourth component is the *outcome* for a parent, either (1) positive or (2) negative. Thus, parents can perceive stress due to their family situation of a child with a developmental disability but can also experience a positive outcome, such as personal growth.

Raising a child with a genetic syndrome is a highly specific child-rearing situation. Research on parenting stress in families with a child with a genetic syndrome has shown that the influence of child characteristics on stress is syndrome-specific (e.g. Fidler et al. 2000) and that children with different genetic syndromes elicit different reactions from their environment (Dykens et al. 2000; Hodapp 1999). Although Hodapp (1999) concludes that for different genetic syndromes behavioral problems are the best predictor of parenting stress, relationships with chronological age have been found for some syndromes as well (Fidler et al. 2000). The only published study of parenting stress in CHARGE syndrome shows that 48% of parents with a child up to 50 months perceive significantly high levels of stress. Parents of children with the syndrome who are also blind report more stress. Neither deafness, nor the number of medical problems has, however, been found to be related to stress. Furthermore high stress levels are related to problems in attachment and parental bonding (Reda and Hartshorne 2008). So far perceptions of parenting stress are only known for parents with very young children with CHARGE syndrome and the relationship of stress with the behavioral phenotype is as yet unknown. This limited knowledge led to the current project.

The first aim was to test the hypothesis put forward by Reda and Hartshorne (2008) that the upbringing of a child with CHARGE syndrome is related to elevated perceived stress levels. This study tested the hypothesis in subjects with a broader age range. The second aim was to test the hypothesis that child characteristics, both behavioral and physical, are related to parenting stress. We tested the specific influence of CHARGE syndrome on the factors: level of adaptive functioning, level of intellectual disability, behavioral problems, ability to speak, auditory and visual problems, deafblindness, gender, and chronological age. In line with research on CHARGE syndrome and several other genetic syndromes (Fidler et al. 2000; Hodapp 1999; Reda and Hartshorne 2008) higher levels of parenting stress were expected to be significantly related to (1) behavioral problems, (2) visual problems, and (3) chronological age. For the other researched factors, this study explored the presence of syndrome-specific relationships with the perceived parenting stress.

Testing these hypotheses is important, since it gives insight into the experience of parents rearing a child with the specific characteristics of CHARGE syndrome. High levels of parenting stress can have severe implications, such as harsh or withdrawn parenting with consequences for child development (Deater-Deckard 2004). Therefore, professionals will need to inform parents about the impact of this syndrome on the entire family system and provide appropriate support in the relevant domains to improve the well-being of the whole family. We have chosen to focus on one specific component of Perry's model (2004) as a possible stressor, namely child characteristics in CHARGE syndrome.

## Method

### Participants

Twenty-two children with CHARGE syndrome (16 boys and 6 girls) and their parents participated. The age of the children<sup>1</sup> ranged from 1.7 to 22.2 years ( $M=11.0$ ,  $SD=5.54$ ). Of the 22 children, 21 had a *CHD7* gene mutation. One child met the criteria of both Blake et al. (1998) and Verloes (2005) for CHARGE syndrome, but genetic screening has not been carried out (yet).

### Procedure

All 55 members of the Dutch CHARGE Parent Support Group were requested through a letter to participate in the current study, and 15 parents agreed to the request. Through collaboration with a Dutch CHARGE-specific outpatient clinic, parents of 11 additional children agreed to participate. Sadly, one child died shortly after his parents had filled out the questionnaires, but they still consented to the use of the data.

Informed consent was obtained for participation in the project. All participants gave written permission for file analysis at the school or day care centre. Parents received the questionnaires by post and were asked to return them through an included pre-paid envelope. Confirmation of the CHARGE diagnosis was either obtained through file analysis or by contacting the medical specialist involved. One child had to be excluded because the CHARGE diagnosis was not clearly supported by the file analysis and the mother did not give permission to contact their medical specialist. In two cases medical specialists were not definite about the presence of CHARGE syndrome; in both cases other genetic syndromes were suspected also. For one child, no *CHD7* gene mutation was found and the criteria of Blake et al. (1998) and Verloes (2005) were not met; this case was excluded from the data-set. Ultimately, data for 22 children were used in the analysis.

### Research Instruments

#### *Measurement of Parenting Stress*

The *Nijmegen Parenting Stress Index-Short* (NPSI-S; De Brock et al. 1992) is an officially translated and adapted version of the Parenting Stress Index by Abidin (1983 in De Brock et al. 1992). It measures parenting stress in families with children from approximately 2 to 13 years. Since level of adaptive functioning of the children did not exceed this level, this instrument was considered appropriate for the purpose. Twenty-five items are scored on a six-point scale. Dutch non-clinical and clinical norms are available for mothers and fathers separately. In this study the non-clinical norm group was used. Internal consistency measured with Cronbach's alpha in the

<sup>1</sup> Besides younger children and adolescents, adults with CHARGE syndrome were included in the project as well. However, as they remain *children of* their parents the term children will be used throughout this article to describe the participants with CHARGE syndrome.

non-clinical and clinical population groups is higher than .91. The NPSI-S shows good criterion validity with accurate prediction of membership of the clinical and non-clinical population. Construct validity is only investigated for the extended version of the instrument: concurrent validity ranges from ‘satisfactory’ to ‘good’ and discriminant validity is considered reasonable (De Brock et al. 1992).

### *Measurement of Child Characteristics*

The *Vineland Screener 0-12 years* (VS 0-12; Van Duijn et al. *in press*) is a Dutch screening instrument adapted from the Vineland Screener by Sparrow et al. (1993). The VS 0-12 measures the level of adaptive functioning of children up to the age of 12 or older people with comparable levels of functioning. An adaptive behavior composite score (90 items) is based on the domains communication, daily living skills, socialization, and motor skills. Parents indicate on a three-point scale whether the child exhibits the particular behavior in everyday life. Good reliability and validity have been established in a normal population. Inter-rater reliability has intra-class correlations for the four domains and adaptive behavior composite between .92–.98, intra-class correlations for test-retest reliability range from .90–.96, and Cronbach’s alphas range from .96–.99 (Van Duijn et al. *in press*). The VS 0-12 years is an expansion of the VS 0-6 years which has proven to have adequate content, construct, and criterion validity (Scholte et al. 2008). A regression formula was developed based upon normal population data to estimate the adaptive level of functioning (Van Duijn et al. 2009).

The Dutch version (Koot and Dekker 2001) of the *Developmental Behavior Checklist-Primary Carer* (DBC-P; Einfeld and Tonge 2002) assesses emotional and behavioral problems in people with an intellectual disability. Parents rate 95 items on a three-point scale about behavior in the past six months. A total behavior problem score is computed together with five subscale scores (disruptive/antisocial behavior, self-absorbed behavior, communication disturbance, anxiety, social relating problems). Intra-class correlations for inter-rater reliability range from .52 to .67 for the total score and the different subscales. Internal consistency (Cronbach’s alphas .66 to .95) and test-retest reliability (intra-class correlations between .76 and .89) are high. Construct and criterion validity are satisfactory (Koot and Dekker 2001). Besides the five subscales the DBC-P has an additional autism screening algorithm which reliably screens for the autistic disorder. Internal consistency is .94 (Einfeld and Tonge 2002). Two other scales with face validity concerning psychiatric conditions are the depression scale and hyperactivity scale. For the depression scale inter-rater reliability and concurrent validity for the depressive disorder have been proven (Tonge and Einfeld 2003). The hyperactivity scale has good construct validity and Cronbach’s alpha is .88 (Einfeld and Tonge 2002).

Information on the expressive communication abilities of the child was gathered through various means. A dichotomous score was coded for speaking/non-speaking. If, according to the parents, the child named or gestured towards some people or things when asked, the child was categorized as ‘speaking’. Parents indicated whether their child had problems with hearing (unable to hear or hears very little) and vision (unable to see or sees very little) on the DBC-P. Children were categorized as being deafblind if parents indicated problems with both hearing and

seeing. All questionnaires have been processed conform the instructions of the official manuals.

## Data Analysis

Based on the VS 0-12 data, the level of adaptive functioning can be calculated using a regression formula that was derived from normal population data. In this study we estimated the level of intellectual disability on the basis of the level of adaptive functioning on the VS 0-12. For children up to 9 years of age, we computed a developmental quotient (DQ) [ $\text{VS 0-12 score} / \text{chronological age} * 100$ ] and classified the level of intellectual disability based upon Došen (2005), see Table 1. Children 10 years and older can no longer obtain a DQ of 100 with the current regression formula. Therefore, we made a classification based upon the developmental level of the older children, see Table 1.

SPSS 14.0 was used for the analyses. Assumptions for Pearson correlations and *t*-tests were met and an alpha of .05 was chosen for all analyses.

## Results

### Parenting Stress

The NPSI-S was filled out for 22 children. This was done by 17 mothers and 1 father. In the remaining four cases, two couples filled it out together and for the other two questionnaires the gender of the respondent was unknown. In these last four cases the norm group for mothers was used. On the NPSI-S the mean raw score was 77.1 ( $SD=30.58$ ), ranging from 25 to 132 (maximum possible score 150).

A large number of parents perceived high levels of stress related to the upbringing of their child. Only 9% scored 'very low' compared to the norm, 4% had stress levels below the mean and 14% scored around the mean of the norm group. Another 14% received a score above the mean. Nearly one-third (27%) experienced high levels of stress and another third (32%) scored within the highest possible category. Compared to the non-clinical norm group, where 10% and 5%, respectively, fell in the high and very high category, this is a very large proportion of the parents.

**Table 1** Classification of intellectual disability based on Došen (2005)

Level of intellectual disability	Developmental quotient	Developmental age
Profound	0–20	<2 years
Severe	20–35	2–4 years
Moderate	35–50	4–7 years
Mild	50–70	7–12 years
None	>70	>12 years

## Child Characteristics

The VS 0-12 was filled out reliably for 20 children. The raw total scores ranged from 18 to 163 (maximum possible score 180). The adaptive level of functioning ranged from 0.2 years to 8.6 years ( $M=4.5$ ,  $SD=3.24$ ). To estimate level of intellectual disability, VS 0-12 scores were transformed as explained in the data analysis section. A wide range of functioning was found. Seven children had a profound ID (32%), one had a severe ID (4%), three had a moderate ID (14%) and four had a mild ID (18%). Five children had no ID (23%). For two children categorizing was not possible (9%), because there were too many missing values on the VS 0-12.

The total problem score on the DBC-P ranged from 3 to 78 (maximum possible score 190). A score above the cut-off point of 46 indicates a substantial number of behavioral problems (Einfeld and Tonge 2002); this was the case for six children (27%).

In Table 2, the findings with regard to the DBC-P subscales are presented. Since the number of items differs between subscales, mean subscale scores were computed to make the scale scores comparable. These scores can range from 0 to 2 and were highest for the hyperactivity subscale followed by the autism screening algorithm (see Table 2).

Furthermore a considerable variation in the behavior of the participants was found. Only 13% of the items were applicable to more than half of the children (i.e. a score of 1 or 2). Behaviors prevalent in 51% to 60% of the children were: aloof, in his/her own world; makes non-speech noises; overly attention-seeking; sleeps too little, disrupted sleep; stubborn, disobedient or uncooperative; underreacts to pain. Five items were prevalent in 61% to 70% of the children: becomes over-excited; poor attention span; has temper tantrums; irritable; noisy or boisterous. The most prevalent behavior was impatience. This was identified in 86% of the children.

Nearly two-thirds of the children (14; 64%) had means of expressing themselves, and thus could be categorized as speaking children, whereas 8 (36%) were non-speaking. Problems with hearing were prevalent among the majority of the children (17; 77%). A smaller group of children (7; 32%) had problems with seeing. All seven children with visual difficulties also had hearing problems and were placed in

**Table 2** Mean subscale scores Developmental Behavior Checklist-Primary Carer ( $N=22$ )

DBC-P subscale	Mean subscale score	Standard deviation
Hyperactivity	.82	.47
Autism screening algorithm	.56	.40
Self-absorbed behavior	.47	.36
Disruptive/antisocial behavior	.46	.27
Social relating problems	.41	.41
Depression	.39	.25
Anxiety	.31	.26
Communication disturbance	.30	.23

the deafblind category (32%). A total of five children had no problems with either hearing or seeing.

### Parenting Stress in Relation to Child Characteristics

Parenting stress was not significantly associated with the level of adaptive functioning of the child with CHARGE syndrome ( $r=-.20, p=.41$ ). To relate level of parenting stress to the level of ID, a dichotomy was made based upon the VS 0-12 results. Children with a profound, severe or moderate ID were grouped together (11 lower functioning children; 55%), as were children with a mild or no ID (9 higher functioning children; 45%). The mean raw NPSI-S score for the lower functioning children was 78.2 ( $SD=31.84$ ) and for the higher functioning children 75.0 ( $SD = 34.02$ ). No significant difference between the mean levels of parenting stress was found,  $t(18)=-.22, p=.83$ .

Parenting stress appeared to be related to certain behavioral problems. All (sub)scales except that of communication disturbances correlated positively with parenting stress. There were significant correlations with four subscales (see Table 3). Higher levels of behavioral problems on the subscales depression ( $R^2=.32$ ), disruptive/antisocial behavior ( $R^2=.19$ ), self-absorbed behavior ( $R^2=.19$ ), and the autism screening algorithm ( $R^2=.19$ ) were related to higher levels of parenting stress. The correlation between parenting stress and the total problem behavior score was not significant, but a  $p$ -value of .05 can be interpreted as a trend ( $R^2=.18$ ). The association between parenting stress and the depression subscale had a large effect size. The associations with the other three significant subscales had medium-sized effects (Cohen 1992).

The stress levels of parents with non-speaking children ( $M=93.4, SD=19.18$ ) were higher than for those with speaking children ( $M=67.9, SD=32.52$ ). Although this difference was not significant at an alpha level of .05, it can be considered a trend in the data ( $t(20)=2.02, p=.06$ ). Parents of hearing children ( $M=80.2, SD=24.51$ ) and those with children who had hearing problems ( $M=76.2, SD=32.77$ ) did not differ in their stress levels,  $t(20)=.25, p=.81$ . Neither was there a difference between parents with children who had good vision ( $M=76.5, SD=27.80$ ) and those

**Table 3** Correlation between raw score Nijmegen Parenting Stress Index-Short and Developmental Behavior Checklist-Primary Carer ( $N=22$ )

DBC-P (sub)scale	Correlation raw NPSI-S score	$p$ value
Depression	.57	.01
Disruptive/antisocial behavior	.44	.04
Self-absorbed behavior	.44	.04
Autism screening algorithm	.44	.04
Total behavior problem score	.42	.05
Social relating problems	.25	.26
Anxiety	.20	.37
Hyperactivity	.15	.51
Communication disturbance	-.13	.55

with children who had problems with seeing ( $M=78.4$ ,  $SD=38.31$ ),  $t(20)=-.13$ ,  $p=.90$ . The children who had visual problems, were all considered deafblind, so this factor was not researched further. The gender of the child had no influence on the NPSI-S scores. Parents of boys ( $M=75.8$ ,  $SD=28.15$ ) experienced similar amounts of stress as parents of girls ( $M=80.7$ ,  $SD=39.12$ ),  $t(20)=-.32$ ,  $p=.75$ . The NPSI-S score was also not related to the chronological age of the child ( $r=.20$ ,  $p=.36$ ).

## Discussion

In line with the first hypothesis it turned out that the upbringing of a child with CHARGE syndrome is related with the experience of high stress levels in two-thirds of the parents. The percentage found was even higher than that reported by Reda and Hartshorne (2008), who investigated only parents of younger children. However, the second hypothesis was only partly corroborated. Specific behavioral problems were related to higher stress levels (i.e. behavior indicative of depression and autistic disorder, disruptive behavior, and self-absorbed behavior, with a trend for the total behavior problem score). The hypothesis that there is an association between parenting stress and chronological age was based upon research into other genetic syndromes (Fidler et al. 2000) and was not confirmed in this study of CHARGE syndrome. Although it lies beyond the reach of this article and study, because of restrictions in analysis-methods with this small number of participants, we assume that the stress parents experience during the lifespan of their child is related to various factors at different ages. Our presumption, based on clinical experience, is that in young children the medical problems with associated surgeries and hospital stays cause a lot of stress for the parents, whereas later in life parents experience more stress because of behavioral problems or worries concerning the development of the child. Although not tested in this article as we looked only into single relationships because of sample size, in our view this would be an important supplementary consideration for future research. The hypothesis that higher stress levels occur in parents with a visually impaired child was also not corroborated although this hypothesis was based upon CHARGE-specific research (Reda and Hartshorne 2008). A possible explanation for this contradictory result could be the difference in defining the visual disability. In the current project this was described as any problem with seeing, whereas Reda and Hartshorne identified a visual disability when no better than moderate visual impairment in the best eye was present. These contradictory results need to be harmonized in future projects to understand the actual influence of visual disability on parenting stress. Besides behavioral problems, no association with parenting stress appeared for the level of adaptive functioning, level of ID, problems in hearing and seeing, deafblindness, gender and chronological age of the child. A trend was found of lower stress levels for parents with speaking children versus those with non-speaking children. Overall the notion of Hodapp (1999) that behavioral problems of children with specific genetic syndromes have the strongest associations with parenting stress was also found to be true for CHARGE syndrome.

As mentioned by Blake et al. (2005) and Vervloed et al. (2006), it is difficult to describe the typical CHARGE person because the characteristics are so highly

variable. Our sample was also heterogeneous with regard to physical and behavioral problems, for example only 13% of the measured behavioral problems were exhibited by more than half of the participants. However, medium to large effects found for several specific behavior patterns show that parenting stress and children's behavioral problems are clearly associated. The shared factor in the participating families is the perception of high levels of stress raising a child with CHARGE syndrome, with these levels being even higher when the children also display behavioral problems.

These findings suggest that professional support for families is an essential part of the assistance needed, and even more so if behavioral problems are present. In such case, parents should get additional support to manage the behavioral problems to lower the stress levels. It must also be emphasized that the child rearing support must be a continuous process, since the stress is not only high among parents with younger children but also among those with older children. Support should thus not be restricted to the turbulent early years of the child's life. As we did not find any significant association between parenting stress and the child factors studied except behavioral problems, professionals should investigate each family individually to determine which factors make the upbringing situation stressful in this particular case. In addition, our experience in an outpatient clinic and the results of Blake et al. (2005) reveal the involvement of many different professionals in the care of these children. The appointment of one professional as a key figure in streamlining all information and as provider of support could relieve parents of this task and promote family well-being. In addition to the care and support for the child with CHARGE syndrome itself, it is of the utmost importance to assist the parents in order to promote the well-being of the whole family system.

However, especially the results on to the relationship between parenting stress and the child characteristics need to be interpreted with caution. A serious problem in many studies, and in this project also, with people with CHARGE syndrome is the small number of participants. This has consequences for the ability to detect a significant effect. According to Cohen (1992) with an alpha of .05, preferred power of .8 and 26 to 28 participants, large effect sizes are needed to get statistically significant outcomes with *t*-tests and Pearson correlations. This poses serious problems for the interpretation and meaning of our and other research results, since it remains unclear whether there truly is no association between the measured child characteristics and parenting stress or whether our sample is simply too small to determine statistically significant effects. Besides this, in the current project participants were distributed unevenly over some categories. For example the number of boys (16) outnumbered the girls (6) and the groups of children with (17) or without (5) hearing problems were also uneven. As it is unclear in which way this may have influenced our results, this is another reason for cautious interpretation.

Another limitation is the use of instruments that are not adapted and normed for this specific population with so many sensory problems. It is possible that the capacities of children with these problems are underestimated by the use of adaptive functioning to categorize the level of ID. However, the use of IQ tests is also problematic, especially for children functioning at the lowest levels with additional disabilities. So far, adaptive functioning may be the best measure we have to give an indication of the abilities of these children. Also, use of the DBC-P could have its

limitations. It could be that children without an ID, exhibit behavioral problems which are not included in the DBC-P. Again the heterogeneity of the sample makes the choice of instruments a complicated issue. However, in our sample only five children were categorized as not having an ID, thus the choice of the DBC-P, based on earlier reports about the level of functioning, seems justified.

In this project we only focused on the relationship between certain child characteristics and the perception of parenting stress. For future projects looking further into these child characteristics is essential. We focused on behavioral problems, but it is also known that there is a heightened risk for psychiatric disorders in CHARGE syndrome, such as autism spectrum disorders, obsessive-compulsive disorder, attention deficit/hyperactivity disorder and Tourette syndrome (Blake et al. 2005; Wachtel et al. 2007). Although the DBC-P describes behaviors characteristic of depression, autistic disorder and hyperactivity, this is not a substitute for an individual descriptive diagnosis. This issue is however not that straightforward; for example, diagnosing autism spectrum disorders in this multi-sensory impaired group is controversial (Hartshorne and Cypher 2004; Johansson et al. 2006; Vervloed et al. 2006). In view of the possible impact of a co-morbid psychiatric disorder, this seems to be an important broadening of the child characteristics measured here. However, as Perry (2004) points out, not only child characteristics influence the perception of parents of the child rearing situation. Focusing more on the differences in the family context, such as differences in resources and support, can give a more comprehensive notion of the complex process that leads to parenting stress in this complex population. In addition, a useful step to include other relevant child and family characteristics would be to first continue with a more qualitative approach by in-depth interviews with parents. In this way specific and new insights can be generated concerning the possible related factors to parenting stress which afterwards can be investigated in a larger CHARGE population with a quantitative approach. Finally, in this study mainly mothers filled out the questionnaires. Studies into parenting stress in both mothers and fathers with a disabled child show contradicting results, but the majority of the studies report comparable stress levels between mothers and fathers (Macias et al. 2007). However, from a clinical perspective, it would be an important additional factor to investigate in this specific population as it can generate valuable knowledge for intervention.

In sum, this study is the first to describe the experience of parents about the upbringing of a child with CHARGE syndrome with a broad age range. The heavy burden of this situation for a substantial part of the parents has become clear, extending Reda and Hartshorne's study (2008). Results regarding the relationship of parental perception and their child's characteristics can be seen as a first exploration of this topic. Perhaps the most important step in research of CHARGE syndrome will be a co-operation between researchers worldwide to be able to collect a large number of children with the syndrome and their families. This will not only resolve the lack of statistical power of studies, but will also help to harmonize measurement methods and research designs, thus raising the validity, reliability, and the generalization of the findings of research with regard to CHARGE syndrome. Although internet surveys among parents in different countries are being carried out already, more active collaboration between researchers in different domains seems necessary.

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