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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7 General conclusions and discussion

INTRODUCTION

The main aim of this study was to expand our knowledge of the behavioural phenotypes of five genetic syndromes associated with intellectual disabilities (ID) and to determine the relationship with perceived parenting stress, in order to improve support through recommendations for clinical practice. The five syndromes are Rett syndrome, CHARGE syndrome, Cornelia de Lange syndrome, Angelman syndrome, and Prader-Willi syndrome. We have reported on the syndromes *separately* in chapters 2 to 5 (only in chapter 6 are two syndromes compared directly). In this final chapter we will present an overview of our findings with regard to the child characteristics and parenting stress, and compare the five syndromes in this respect. Thereafter, a critical reflection of the present study is given and suggestions for future research are provided. The chapter ends with implications for clinical practice.

OVERVIEW OF FIVE GENETIC SYNDROMES

For all five syndromes the behavioural characteristics were investigated by the same assessment instruments, but for each syndrome specific emphasis has been placed upon *different* child characteristics in chapters 2 to 6. Subsequently, a description is provided of the behavioural phenotype of the same aspects for all five syndromes, based upon the shared data presented in chapters 2 to 6, i.e. on adaptive functioning and level of ID, the presence of the autistic disorder, and behavioural problems. The similarities and differences between the syndromes will be discussed and remarkable findings per syndrome will be highlighted as far as these were not already mentioned in chapters 2 to 6. Finally the relationships between the behavioural phenotype and the perceived parenting stress will be compared between the genetic syndromes investigated.

The behavioural phenotypes of five genetic syndromes

A summary of the different child characteristics per syndrome is given in Table 7.1. The behavioural phenotypes for the genetic syndromes are compared, although this comparison is somewhat hampered by the uneven age range and gender composition of the samples and therefore must be viewed with some reserve.

With regard to the level of **adaptive functioning**, a clear distinction emerges for the maximum level reached. Those with Rett syndrome or Angelman syndrome reach adaptive developmental ages of two to three years. Those with CHARGE syndrome, Cornelia de Lange syndrome, or Prader-Willi syndrome have a much higher maximum level, i.e. adaptive developmental ages of eight to nine years.

By estimating the level of **intellectual disability**, based on the adaptive functioning and taking chronological age into account (see e.g. chapter 4), a similar distinction was found. All individuals with Rett syndrome or Angelman syndrome have a severe to profound ID. In the other three syndromes there is more variation and all levels of functioning (profound ID to no/mild ID) are present. However, in these syndromes the distribution of ID level is syndrome-specific. For CHARGE syndrome the lowest (severe to profound ID) and highest levels of functioning (no to mild ID) are equally present. Thus, for children with CHARGE syndrome the level of functioning is hard to predict. Persons with Cornelia de Lange syndrome mostly function in the severe to profound disabled range, although higher functioning individuals are also present. The majority of the children with Prader-Willi syndrome have no to a mild ID, but a proportion functions at the lower levels.

In other studies of both Rett syndrome and Angelman syndrome higher levels of functioning have occasionally been found (Demeter, 2000; Duker, Van Driel, & Van de Bercken, 2002; Peters et al., 2004; Thomson, Glasson, & Bittles, 2006), but these levels of abilities seem exceptional and were not seen in our study. For CHARGE syndrome a broad range of functioning has been described (Johansson et al., 2006; Salem-Hartshorne & Jacob, 2005; Smith, Nichols, Issekutz, & Blake, 2005), but the current study indicates that there is a *substantial* percentage that functions in the (near) normal range.

In the literature on the general population of people with ID, the prevalence rate of the **autistic disorder** is linked to the level of ID, but the exact prevalence is unclear because rates differ considerably between studies. Deb and Prasad (1994) found that 37% of the children with severe to profound ID had the autistic disorder, 16% of the children with moderate ID, and 8% of the children with mild ID. De Bildt, Sytema, Kraijer, and Minderaa (2005) found a prevalence rate of pervasive developmental disorders (including the autistic disorder and PDD-NOS) of 26% for children with moderate to profound ID and 9% for children with mild ID. The global outcome is that the highest prevalence rates

for the autistic disorder are found at the lower end of the ID spectrum (De Bildt et al., 2005).

In the current study we screened for the autistic disorder with the Developmental Behaviour Checklist - Autism Screening Algorithm (DBC-ASA; Einfeld & Tonge, 2002) and the Diagnostic Interview for Social and Communication Disorders - 10th Revision (DISCO-10; Wing, 1999). Table 7.1 shows the percentage of agreement (autistic disorder present or not present) and disagreement on classification (uncertain) between the instruments. The two instruments suggest that the autistic disorder is present in somewhat more than a third of the females with Rett syndrome and this is a similar to somewhat higher proportion expected for persons with this level of ID (Deb & Prasad, 1994; De Bildt et al., 2005). For a discussion on the controversial issue to classify the autistic disorder in females with Rett syndrome, see chapter 2. When a child has Angelman syndrome, the autistic disorder is suspected to be present in two-thirds of the individuals. This is a much higher proportion than in the general population of children with severe to profound ID. For the three other syndromes, the comparison is more complicated since a broad range of ID levels is thereby present. In Cornelia de Lange syndrome in more than half of the cases a co-morbid autistic disorder is suspected. This is a higher proportion than expected, even if the highest prevalence rates, related to severe and profound ID, are taken into account. For CHARGE syndrome and Prader-Willi syndrome percentages are considerably lower, although still substantial.

In all of the syndromes investigated, with the exception of Rett syndrome, there seems to be an increased risk of a co-morbid autistic disorder given the level of functioning within the syndromes. However, these figures for the presence of the autistic disorder need to be considered cautiously. In the current study screening instruments were used and thus only estimates can be given; individual assessment should always follow the screening to obtain a diagnosis. This step is certainly needed to be definite about the risk for co-morbidity with the autistic disorder in these genetic syndromes.

A population study of individuals with ID revealed that 41% had a severe amount of **behavioural problems**, i.e. measured as the percentage of individuals with scores in the clinical range (Einfeld & Tonge, 1996). In our study, a higher percentage of individuals with Cornelia de Lange syndrome and Angelman syndrome exhibited a severe amount of behavioural problems (see Table 7.1). A lower percentage of persons with Rett

Table 7.1 *Overview of child characteristics per syndrome based upon chapters 2 to 6*

		RS	CS	CdLS	AS	PWS
N		52	22	37	24	23
Gender	Male	-	16	21	11	10
	Female	52	6	16	13	13
Age in years	range	2 - 49	1 - 22	1 - 46	2 - 12	2 - 12
	(*M; SD*)	(16.5; 11.8)	(11.0; 5.5)	(18.1; 13.0)	(8.6; 3.1)	(7.3; 3.2)
Adaptive functioning	range	2 - 28	3 - 103	4 - 112	0 - 33	3 - 101
(in months)	(*M; SD*)	(7.6; 4.4) [a]	(53.5; 38.9) [b]	(26.4; 26.1) [c]	(9.4; 10.3) [b]	(53.6; 24.5) [b]
Intellectual disability [d]	severe-profound	100%	40%	68%	100%	26%
	moderate	-	15%	6%	-	9%
	no-mild	-	45%	16% [c]	-	65%
Autistic disorder [e]		39%	36% [g]	54%	67%	22%
	uncertain [f]	23%	9%	16%	25%	26%
	not present	38%	55% [g]	30%	8%	52%
Severe behavioural problems [h]		25%	27%	47%	54%	35%

RS=Rett syndrome; CS=CHARGE syndrome; CdLS=Cornelia de Lange syndrome; AS=Angelman syndrome; PWS=Prader-Willi syndrome.

[a]Based on Vineland Screener 0-6;

[b]Based on Vineland Screener 0-12;

[c]Based on VABS.

[d]For children aged 1 to 9 years a developmental quotient was calculated based upon relevant version of Vineland Screener. For children aged 10 years and older the developmental age was used to classify the level of intellectual disability, see chapter 4 for more explanation.

[e]Based upon DBC-ASA and DISCO-10;

[f]The two instruments disagree about classification of autistic disorder;

[g]For one individual based upon only DBC-ASA.

[h]Clinical number of behavioural problems based on cut-off DBC.

syndrome, CHARGE syndrome, or Prader-Willi syndrome showed clinically significant behavioural problems in comparison to the general ID population. Thus, the presence of Angelman syndrome or Cornelia de Lange syndrome can be seen as factors that heighten the risk of severe behavioural problems. To get more insight into the behavioural problems that are specific for each syndrome, in Table 7.2 problems are tabulated which are prevalent in more than 70% of the individuals (i.e. receive a rating that the behaviour is 'somewhat or sometimes true' or 'very true or often true'). Those with CHARGE syndrome show the most variation in behavioural problems; only one behavioural problem (impatience) is present in more than 70% of the individuals. In Rett syndrome, Cornelia de Lange syndrome, and Prader-Willi syndrome six to eight behavioural problems are highly prevalent. Some of these behaviours are highly prevalent in one of the other syndromes as well, but in all three syndromes some *specific* problems appear highly prevalent. Children with Angelman syndrome are the most alike as far as behavioural problems are concerned. For them, 15 behaviours are prevalent in the majority of the children; eight of these behaviours are not present in the majority of any of the other four syndromes, i.e. can be marked as a unique characteristic of the syndrome.

Because the behavioural problems were measured with the same instrument, and the ID level is taken into account, valid between-syndrome comparisons can be made. Of the two syndromes associated with severe to profound ID, those with Angelman syndrome are more alike in their behavioural problems than individuals with Rett syndrome. For the three syndromes with mixed levels of functioning, those with CHARGE syndrome are much more varied in their behavioural problems than those with Cornelia de Lange or Prader-Willi syndrome. Overall, the behavioural phenotype is most distinct for Angelman syndrome, whereas in CHARGE syndrome one can barely speak of a behavioural phenotype.

Parenting stress and associated child characteristics in five genetic syndromes

The level of parenting stress that is perceived by parents with a child with one of the syndromes is depicted in Table 7.3. Parenting stress is rated as high when the scores fall into the two highest categories of the normal population norm group, covering 15% of parents in the general population who report stress related to the child-rearing situation. Raising a child with one of the genetic syndromes investigated is a substantial risk factor

Table 7.2 *Developmental Behaviour Checklist items that are applicable to more than 70% of the participants per syndrome*

Rett syndrome (*N* = 52; 2 - 49 years)	
Item	**%**
Repeated movements of hand, body, head, or face	92
Poor attention span	87
Laughs or giggles for no obvious reason	81
Makes non-speech noises	79
Underreacts to pain	75
Aloof, in her own world	73

Angelman syndrome (*N* = 24; 2 - 12 years)	
Item	**%**
Chews or mouths objects, or body parts	96
Poor attention span	96
Easily distracted from his/her task	92
Makes non-speech noises	92
Sleeps too little, disrupted sleep	92
Overactive	88
Poor sense of danger	88
Unusual body movements, posture, or way of walking	83
Likes to hold or play with unusual object, overly fascinated with something e.g. water	79
Repeated movements of hand, body, head, or face	79
Becomes over-excited	75
Eats non-food items	75
Impatient	75
Stubborn, disobedient, or unco-operative	75
Unrealistically happy or elated	75

Cornelia de Lange syndrome (*N* = 37; 1- 46 years)	
Item	**%**
Prefers to do things on his/her own, tends to be a loner	81
Mood changes rapidly for no apparent reason	78
Aloof, in his/her own world	75
Makes non-speech noises	75
Poor attention span	75
Avoids eye contact	72

Prader-Willi syndrome (*N* = 23; 2 - 12 years)	
Item	**%**
Arranges objects or routine in a strict order	91
Easily distracted from his/her task	87
Impatient	83
Poor sense of danger	83
Upset over small changes in routine or environment	83
Easily led by others	74
Scratches or picks at his/her skin	74
Stubborn, disobedient, or unco-operative	74

CHARGE syndrome (*N* = 22; 1 - 22 years)	
Item	**%**
Impatient	86

Note. Items that are applicable are given a score 1 or 2. Items in italics are 'unique' in being prevalent in more than 70% of the syndrome compared to the other syndromes.

for experiencing high levels of parenting stress. Parents with a child with Prader-Willi syndrome report less stress than parents with a child with one of the four other syndromes, although the percentage who perceive high stress is still higher than in the normal population. However, in addition to underlining this risk factor, it should be mentioned that there are also a lot of parents with a child with one of the syndromes who do not perceive the child-rearing situation as highly stressful.

Having investigated the (sometimes problematic) characteristics of the children, it is a logical step to assume that there are relationships between child characteristics and parenting stress. Hodapp (1999) has suggested that the degree of parenting stress in genetic syndromes is best predicted by the child's behavioural problems. Table 7.3 shows that for the various syndromes different child characteristics relate to parenting stress. For example for parents with a child with Cornelia de Lange syndrome, stress is significantly higher when the child functions at a lower level, whereas in CHARGE syndrome the level of functioning is not related to parenting stress. Although in both syndromes considerable variation in level of functioning exists, it depends on the syndrome whether this factor relates to parenting stress or not. This suggests that relationships between child characteristics and parenting stress are syndrome-specific.

CRITICAL REFLECTIONS AND DIRECTIONS FOR FUTURE RESEARCH

Some limitations became apparent in the current research project, that can be used to improve future studies. In this study the *relationship* between child characteristics and parenting stress was investigated by means of a cross-sectional descriptive design. Carrying out detailed descriptive research is as important as searching for causality in stress research (Lazarus, 2000). However, knowledge about causes can lead to better well-aimed interventions. In the current study, Perry's model (2004) (see Figure 1.1) was used as a framework wherein child characteristics are depicted as stressors with parenting stress as a negative outcome, and thereby suggest causality. But using this model to investigate these variables does not imply that conclusions about *relationships* in the current study can be extended to conclusions about *causality* without direct testing. For children with ID in general, the issue of causality of parenting stress and child characteristics is as yet not resolved. In some studies child characteristics, often behavioural problems, are found to cause parenting stress, whereas a substantial number of studies has reported a bi-

Table 7.3 *Overview of parenting stress per syndrome based upon chapters 2 to 6*

	RS	CS	CdLS	AS	PWS
N	24 [a]	22	37	24	23
High parenting stress [b]	46%	59%	51%	58%	26%
Child characteristics tested on relationship with parenting stress	- age -adaptive functioning - behavioural problems - autistic disorder - Rett-specific behaviours	-gender - age -adaptive functioning - intellectual disability - behavioural problems - ability to speak - auditory and visual problems - deafblindness	- gender - age - intellectual disability - behavioural problems - autistic disorder - severity of physical problems - gene mutation type	- gender - age - behavioural problems	- gender - age - intellectual disability - behavioural problems
Relationships between child characteristics and parenting stress	more total behavioural problems and in subscales self-absorbed, autism, social relating, anxiety and disruptive behaviour, more Rett-specific general mood problems → higher parenting stress	more behavioural problems in subscales depression, autism, self-absorbed, and disruptive behaviour → higher parenting stress	older age, more severe intellectual disability, more behavioural problems, presence autistic disorder → higher parenting stress	no relation between measured child characteristics and parenting stress	no relation between measured child characteristics and parenting stress

RS=Rett syndrome; CS=CHARGE syndrome; CdLS=Cornelia de Lange syndrome; AS=Angelman syndrome; PWS=Prader-Willi syndrome.
[a]Based upon a subgroup of females with Rett syndrome, 24 children aged 2 to 18 years, see chapter 4.
[b]Scores falling in the two highest norm categories of normal population on Nijmegen Parenting Stress Index - Short.

directional effect (Hassall & Rose, 2005; Hastings & Beck, 2004; Hastings, Daley, Burns, & Beck, 2006; Olsson, 2008). Therefore, longitudinal studies in the field of genetic syndromes are needed to test causal directions in these specific populations.

The behavioural phenotype of five genetic syndromes was described (see Table 7.1 and Table 7.2). If the definition of Dykens (1995) for behavioural phenotypes is used, the statement that a particular behavioural problem belongs to the behavioural phenotype requires an adequate control group (e.g. on level of functioning and age) (Einfeld & Hall, 1994). We were able to compare syndromes with comparable levels of functioning (i.e. Rett and Angelman syndrome; CHARGE, Cornelia de Lange and Prader-Willi syndrome) and as such statements about syndrome-specific behaviour can be made. However, the individuals were not directly matched for level of functioning, age, and gender and those with a genetic syndrome were compared mutually. In future studies a matched control group with a non-specific cause for ID is required and more in-depth comparisons between the five syndromes are needed to reconfirm the statements about the behavioural phenotypes that became evident in the current study.

In Perry's model (2004; see Figure 1.1) our focus was on child characteristics as stressors because these are the core and distinguishing features of children with genetic syndromes. We have chosen to investigate child characteristics that were relevant because they can be highly disturbing, i.e. behavioural problems, autistic disorder symptoms, and low levels of independence. In future studies it is essential to broaden the child characteristics measured. Although numerous child characteristics can be mentioned, a few syndrome-specific recommendations are provided. In both Rett syndrome and CHARGE syndrome physical disabilities are often present and can be very severe. Measurement of the relationship between physical characteristics and parenting stress could shed light on the impact of the child's physical problems on the upbringing situation. In Cornelia de Lange syndrome the autistic disorder is highly prevalent. Parents with a child with Cornelia de Lange syndrome and the autistic disorder perceive more parenting stress. To improve support it would be helpful to investigate which specific aspects of the autistic disorder are perceived as stressful. In Angelman syndrome an important child characteristic to include in studies would be a detailed measure of non-verbal communication abilities. Having a child who does not talk and is also unable to communicate non-verbally might be a strong influencing factor; this is highly relevant for the Angelman syndrome since non-verbal abilities differ substantially in this population.

In Prader-Willi syndrome it would be useful to measure the intensity of support that is needed to manage the child's eating habits and to determine the extent to which this poses a burden for the child-rearing situation.

Besides expanding and specifying child characteristics, research will also improve by including additional elements of Perry's model (2004) to gain more insight into risk as well as protective factors. Other stressors than characteristics of the child with the genetic syndrome can be influential. Some of these stressors are specific for families with a child with ID (e.g. additional expenses for adapting the home) whereas others can appear in every family (e.g. unemployment) and both types should be investigated in future studies. The inclusion of measures for mediating resources and (in)formal support (see Figure 1.1) will provide a more comprehensive description of the stress process. Measurement of parental coping strategies and the amount and type of formal and informal support seem thereby to be the minimum essentials needed to gain insight into stress protective factors. Although our focus was on negative outcomes for parents (i.e. parenting stress), positive outcomes should be measured simultaneously since parents can also experience positive aspects of having a child with a genetic syndrome. If more measurements of all aspects of Perry's model are included, this will do more justice to the reality of the child-rearing situation in these families. We follow Olsson's (2008) view that in future studies it is important to focus on the *processes* that lead to different outcomes in families with a child with a certain genetic syndrome. Why do some families adapt well to their specific situation and others do not? There is still a lot to discover about causality and influencing risk and protective factors by means of research in families with genetic syndromes.

Limitations are further posed by the size and recruitment of the samples. The numbers of participants included in the analyses (see chapters 2 to 6) were, respectively, 52, 24, 22, 37, 24, and 23 families. Given the rarity of the five genetic syndromes and the size of the Netherlands population, these are acceptable figures. However, the small sample sizes result in a lack of statistical power. This poses serious threats for the conclusions that can be drawn from this study; this is a challenge for a lot of other studies into genetic syndromes as well. It remains thus uncertain whether there truly is no effect or whether our group was simply too small to detect it. Therefore, closer international collaboration between researchers investigating genetic syndromes is needed to expand sample sizes. Although worldwide data bases are already used for research into gene mutations, e.g. in Rett syndrome, a comparable initiative is needed in the behavioural

sciences. There are research instruments that have been translated world-wide and that have clearly been proven to be useful within the ID population, such as the Developmental Behaviour Checklist (Einfeld & Tonge, 2002). Solely by using such instruments and sharing data world-wide will behavioural studies of genetic syndromes be able to make a big step forward.

Recruitment of the participants took place via the various Dutch Parent Support Groups. Members of such support groups have been characterised as being highly motivated and of middle to high socio-economic status (Dykens, 1999). Only a proportion of the members of the support groups participated in our project. The problem is that one cannot know the representativeness of members of a support group, in particular the self-selected sample of the support group. In CHARGE syndrome we collaborated with a specialised outpatient clinic to gather more participants. Again, it is not known what specific characteristics these families have, but it is highly likely that there is also a selection of people who visit such a clinic. The investigated genetic syndromes are rare and both ways of recruiting people induce uncertainty about the representativeness of the sample. Using all available tracks simultaneously, parent support groups, specialised clinics, organisations and institutions for people with ID, seems the best way to gather as many participants as possible, because the perfect way for recruitment in this field simply does not exist (Finegan, 1998).

CLINICAL IMPLICATIONS

Based upon the results of the present study, the following recommendations for clinical practice can be given. First, the general implications applicable to all the five syndromes will be presented, then some additional syndrome-specific recommendations will be discussed (see also chapters 2 to 6).

General recommendations

Because the behavioural phenotypes of the syndromes investigated in the present study vary considerably, it is important that professionals provide parents of a child with a genetic syndrome with a detailed description of the behavioural strengths and weaknesses that are associated with their child's syndrome. Specialised psychoeducation can show that associated behavioural problems are not displayed on purpose by the child and parents are

not to blame for the presence of these behaviours. Parents can then also try to anticipate the child's behaviour and developmental abilities (Finegan, 1998; Skuse, 2000).

Awareness of the seemingly high prevalence of a co-morbid autistic disorder in at least a proportion of the syndromes is essential because this has a big impact on those with ID (De Bildt, 2003; Van Berckelaer-Onnes, 1996). Although we only *screened* for the autistic disorder, our results nevertheless suggest that in all the syndromes investigated, except Rett syndrome, there is a heightened prevalence of autistic disorder symptoms compared to those with the same level of functioning without a genetic syndrome. However, there are ongoing discussions whether the autistic disorder should be classified in people with genetic syndromes or whether the symptoms in genetic syndromes have different profiles and thus should be labelled as autistic traits (see Moss & Howlin, 2009, for a detailed discussion). Regardless of the outcome of this discussion, the advice for clinicians will be the same: Individuals with a genetic syndrome and a co-morbid autistic disorder (*or* autistic traits) should be given the same support and interventions as people with ID and the autistic disorder with additional adaptations needed per genetic syndrome. Kraijer (2004) provides three core strategies for people with autism spectrum disorders and ID. First, structure and predictability are essential in daily life routine. Second, the demands that are placed upon people with this double diagnosis should be adapted to their often disharmonic functioning. Third, alternative ways of communication are necessary. The quality of life for this population can be increased when augmentative communication is attuned at the right level of sense-making (Noens & Van Berckelaer-Onnes, 2004). Adaptation of the environment is thus essential. Clinicians should thereby integrate the specific approaches for people with these three diagnoses (i.e. a genetic syndrome, ID, and the autistic disorder) and tune into the individual's need for support.

Professionals involved in the support of a child with a genetic syndrome should not only focus on the child's needs, but also on the family system. Although there are parents who do not perceive the child-rearing situation as stressful, our study also shows that there are many parents who perceive high levels of parenting stress. Parenting stress can have severe negative consequences for both parents and child (e.g. Deater-Deckard, 2004; Oelofsen & Richardson, 2006; Pazcowski & Baker, 2007; Singer, 2006). Therefore, professionals should give family assessment a prominent place in order to detect highly distressed parents for whom support is needed. If this is the case, several steps can be taken. First, it is important to provide parents with information concerning their child's

genetic syndrome (Bass, 1990). Providing information on the child's strengths and weaknesses in behaviour has been mentioned before, but information should also be provided on e.g. the aetiology, medication, and possible therapies. A parent support group for the particular syndrome can be a very important additional source of information for parents. Professionals therefore need to inform parents about the existence of such groups and encourage membership. This is especially important since parent support groups not only provide information, but members can provide emotional support for each other. Sharing experiences with someone who experiences similar problems (i.e. other parents) can give a sense of belonging and enhances the caregivers abilities to cope with stress. Parent support groups thus can give parents more confidence concerning their caring tasks and as such play an important role in empowerment of parents (Bass, 1990). Second, in three of the currently investigated syndromes parenting stress is higher when children exhibit behavioural problems. Parents with a child with Rett syndrome, CHARGE syndrome, and Cornelia de Lange syndrome with high levels of behavioural problems need support to manage these behavioural problems which in turn will reduce parenting stress (Hastings & Beck, 2004). Third, parents should be supported to limit the levels of stress and highly distressed parents should be offered stress management strategies which can be helpful in coping with different situations throughout the upbringing process. This is needed, as in all five syndromes parenting stress did not reduce when the child grew older. Having a child with a genetic syndrome remains stressful and this emphasizes the need for the family support to be a continuous process in order to provide information and advice at different stages of life.

Many different disciplines are involved in the care of children with genetic syndromes associated with ID, because both medical and behavioural problems often exist. A lot of the children participating in the current project were seen not only by many different experts but also by a lot of different disciplines. This corresponds to, for example, the finding that in CHARGE syndrome on average 17 different professionals were seen on a regular basis in caring for these children (Hartshorne, 1993 in Hartshorne & Hartshorne, 1998). It can be distressing for parents to obtain (sometimes contrasting) information from so many experts. For some parents it takes a vast amount of time to manage all information and appointments for their child and this is often experienced as highly distressing. It is important that these families are supported by one professional

who becomes a key figure in streamlining and, most importantly, integrating all of the information and thus relieves some of the heavy burden these parents face.

Additional syndrome-specific recommendations

Rett syndrome There is discussion concerning the placement of Rett syndrome under the pervasive developmental disorder section in the major classification systems for mental and health disorders (see chapter 2; Wulffaert, Van Berckelaer-Onnes, & Scholte, 2009). Our results suggest that some females with Rett syndrome have an additional autistic disorder and professionals need to be alert for the presence or absence of this co-occurrence. The prevalence of this co-morbid disorder is in line with studies of individuals with severe to profound ID without Rett syndrome. We underline this co-morbidity explicitly as in the major classification systems the presence of Rett syndrome precludes the possibility of a co-morbid classification of the autistic disorder. For the next version of the Diagnostic and Statistical Manual of Mental Disorders, the DSM-V, it is already proposed to remove Rett syndrome from the pervasive developmental disorders (American Psychiatric Association, 2010).

As mentioned before, parents need support to manage the behavioural problems of their child with Rett syndrome. Additionally, we adopt the advice by Sarimski (2003) and Laurvick, Msall et al. (2006) that support should also focus on the challenges caused by the physical disabilities in Rett syndrome. Furthermore, in these studies a positive impact on the family system was found when mothers had time for their own activities besides caretaking, such as having work outside the house and free time, which thus should be encouraged.

CHARGE syndrome Parents and professionals working with children with CHARGE syndrome need to be aware of the variability in level of functioning in the syndrome. A substantial proportion of these individuals function in the normal to near normal range. In the early years of family life the focus lies mainly on the child's medical problems, and understimulation of the cognitive development is a substantial risk. Given the broad range of abilities, the cognitive and behavioural development of these individuals should be given attention as soon as possible after medical problems are stable or under control. The autistic disorder is suspected to be present in a substantial proportion of those with CHARGE syndrome. In addition, a lot of them have sensory deficits which severely affect development. The combination of these problems, which both have an

impact on the perception of daily life, makes communication an important domain for early intervention in this syndrome.

Cornelia de Lange syndrome Parents of children with Cornelia de Lange syndrome and professionals working with them should be aware of the highly variable behavioural and physical phenotypes. When working with families with a young child, professionals should know that the physical phenotype is not to be used as a prognostic factor for the level of functioning of the children. Given the broad range of functioning of individuals with the syndrome, it is important to monitor the development of young children closely and offer stimulation adjusted to the level of functioning. Severe behavioural problems are present in a large proportion of those with Cornelia de Lange syndrome and parents should obtain professional support to manage and/or reduce these problems.

Half of the parents with a child with Cornelia de Lange syndrome experience high stress levels. Risk factors in families with a child with Cornelia de Lange syndrome are a low level of functioning of the child, high level of behavioural problems, the presence of the autistic disorder and older age of the child. Professionals need to be alert when these risk factors are present in order to provide early support and prevent problems from getting worse.

Angelman syndrome In Angelman syndrome severe behavioural problems are also present in a large proportion of the children. Their parents should obtain professional support to manage and/or reduce these problems. Parenting stress is high in a large proportion of the families with a child with Angelman syndrome. In our study no specific child characteristics in Angelman syndrome were found that were related to parenting stress. However, we provide some hypotheses in which domains families could receive support. We hypothesized that support should focus on optimising the communication abilities of the child, which we discussed at the national family day 2009 of the Dutch Angelman Parent Support Group. Parents agreed that poor communication abilities of their child were a source of stress, and some commented that they were even more concerned whether professionals could understand their child as well as they themselves did. Another target for intervention in order to reduce parenting stress is to focus on the sleep problems suffered by the majority of these children, which also was discussed at the national family day. These suggestions give rise to further investigations. The stress

process in Angelman syndrome needs to be unravelled further to give more specific advice.

Prader-Willi syndrome The majority of the children with Prader-Willi syndrome have a mild to no ID. However, there is also a substantial number of children who function at lower levels. It is therefore important to investigate the abilities at an early age in order to choose the most appropriate level of schooling for these children, in order to prevent under- as well as overstimulation. Most parents with a child with Prader-Willi syndrome perceive a somewhat heightened level of parenting stress (i.e. above the mean of the normal population norm group), but a quarter perceives high levels of parenting stress. However, in our project no specific child characteristics were found that were related to parenting stress. Further investigations are needed to shed more light on this issue and provide more syndrome-specific recommendations for support.

Final remark

To conclude, we studied *groups* of children with genetic syndromes and their families. We emphasize that therefore only general guidelines can be given. Genetic determinism should thereby be avoided; the presence of a genetic syndrome is only a predisposition for certain outcomes. Individuals with the same syndrome differ from each other in e.g. behavioural characteristics (Hodapp & Dykens, 2004, 2009) and the families differ from each other. Parents value professionals who see the individuality and uniqueness of a family (Lärka Paulin, Bernehäll Claesson, & Brodin, 2001 in Olsson, 2008). Support for families with a child with a genetic syndrome should therefore be based on scientific knowledge, but comprise individual assessment to get insight into the challenges and influencing factors in *that* particular family. By expanding specific knowledge on these children and their families, it will become possible to formulate syndrome-specific guidelines for diagnostics and treatment of both medical and behavioural aspects throughout the lifetime, such as Kline et al. (2007) already did for persons with Cornelia de Lange syndrome. This will improve the care and support that people with genetic syndromes associated with ID receive.