Growing up with Down syndrome
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Chapter 8

Summary
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This thesis titled ‘Growing up with Down syndrome - The developing child and its parents’ focuses in the first part on the developing child with Down syndrome (DS), and in the second part on the parents raising a child with DS.

PART 1 – THE DEVELOPING CHILD WITH DS

The developing child with DS was studied from the perspective of a randomized controlled trial concerning thyroid hormone treatment (T4) in the first two years of life and a follow-up study at the age of 10.7 years. The effect of early T4 treatment was studied since newborns with DS, as a group, show signs of mild hypothyroidism. Particularly early in life optimal thyroid hormone levels are important for healthy brain maturation and, to a lesser extent, for physical growth. This trial included children who would otherwise not be treated, i.e. children with normal or at most subclinical thyroid hormone concentrations. Indeed, this trial revealed that T4 treatment during the first two years of life resulted in somewhat better motor development and growth. It was hard to predict, however, whether these benefits at the age of 2 years would result in benefits later in life. Therefore, a follow-up study was needed to establish the long-term effect of T4 treatment in early life.

Chapter 2 describes the results of this follow-up study, which consisted of a single visit at the age of 10.7 years. T4 treatment in early life did not result in any significant long-term developmental benefit. Concerning growth, however, T4 treated children did show some advantage at the age of 10.7 years, particularly those with signs of neonatal subclinical hypothyroidism. This implies that early T4 treatment with the goal of stimulating long term development is not recommended in children with normal thyroid hormone concentrations. It is still possible that children with more severe subclinical hypothyroidism than we could study, would profit from early T4 treatment. This is an issue that is not only relevant for DS, but for all children with subclinical hypothyroidism in early life. High quality research on early T4 treatment in this group is needed. Although increasing growth is in itself not a compelling reason to treat children with T4 in early life, it does suggest that thyroid hormones play an important role in early growth in children with DS.

Chapter 3 describes in more detail the developmental outcomes in our unique cohort of children with DS. At the age of 10.7 years, there was a wide variability in intelligence, adaptive functioning and motor skills across children with DS. Still, as a group, particular deficits and relative strengths in adaptive functioning and motor skills were found, as suggested by previous literature. Yet, where literature generally assumes that there is a single behavioral phenotype in DS, we found considerable differences in the profiles of children who were relatively high functioning versus those who were relatively low functioning. This was particularly so for adaptive functioning, where communication was one of the relatively strong skills in the high functioning subgroup, but the weakest skill in the low functioning subgroup. We have also determined the relation between early life characteristics and developmental outcomes at the age of 10 years. Early developmental outcomes were the
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most important predictor for the level of functioning at age 10 years, while male gender and the occurrence of infantile spasms refined this prediction by predicting poorer functioning later in life. Motor skills appeared to be less well predicted by early life characteristics. Findings in this study can be used as reference for developmental outcomes at group level, and to provide some prognosis of the developmental trajectory of a child. This can be useful to guide parents or to plan for interventions or education. This should, however, be done with caution, since the majority of variation remains unexplained.

PART 2 – PARENTING A CHILD WITH DS

The second part of this thesis focuses on parents of children with DS who participated in the thyroid hormone trial and in the follow-up study. Their experiences were described in terms of health related quality of life (HRQoL) and family functioning, and describe distress and everyday problems as reported on a psychosocial screening instrument. Unlike many previous studies, we included the perspectives of fathers, and describe parents of children within relatively narrow age ranges, since the parenting experience may change considerably with age of the child.

Chapter 4 describes predictors of parental HRQoL domains that were found to be lower in parents of 6 to 8 year olds with DS as compared with parents of children without DS: cognitive functioning, social functioning, daily activities, and vitality. We found that unfavorable outcomes were predicted by a range of variables, of which psychosocial variables appeared as the most consistent and powerful predictors. These considered mainly social support, the quality of the partner relation and time demands.

Chapter 5 focuses on parental HRQoL and family functioning as reported by mothers and fathers of 11 to 13 year olds with DS. Parental HRQoL showed few statistically significant differences from that of control parents. Judging by the effect sizes for the domains that showed a tendency towards lower HRQoL, however, there appeared to be relevant HRQoL issues, which were largely in line with the findings in chapter 4. In parents (mostly mothers) who completed the questionnaires both at child’s age 6 to 8 years and 11 to 13 years we found no significant change over time. Family functioning outcomes were clearly unfavorable in parents of children with DS. Particularly the domains partner relation and social network were unfavorable in both mothers and fathers, while for fathers this was also the case for domains concerning parenting.

Chapter 6 describes outcomes of a frequently used psychosocial screening instrument, the Distress Thermometer for Parents. This instrument assesses the burden that parents experience (on a scale from 0 to 10) and the occurrence of 34 common everyday problems. Mothers and fathers of 11 to 13 year old children with DS were compared with mothers and fathers of 11 to 13 year old children without DS. Clinical distress was not more frequent among parents of children with DS, and mothers of children with DS did not report more everyday problems than control mothers. Fathers, however, reported more problems than controls across almost all problem domains and more frequently wished to consult
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a professional about their situation. This suggests that a high number of problems was a better indication for the need to consult a professional than the distress score.

Taken together, the outcomes in chapters 4 to 6 seemed contradictory in some respects. For instance the consequences in terms of HRQoL were subtle compared with the family functioning issues, as described in chapter 5. Also, there was an apparent discrepancy between normal distress scores and high number of problems reported by fathers in chapter 6. In the general discussion of this thesis in chapter 7 it is hypothesized that these apparent contradictions reflect a response shift. A response shift is thought to result from a change in expectations, values and standards, which has been reported before in parents of children with DS. On more factual questions concerning functioning, parents appear to report less favorable outcomes than controls, while questions concerning the subjective evaluation of their functioning reveal little difference from controls. Important themes in the functioning of parents concerned social functioning, partner relation, vitality, participation in activities, and emotional functioning. Fathers of young adolescents with DS appear to experience problems with parenting.

Professionals caring for these families need to be aware of these important themes, and make an effort to address these. A balanced perspective is needed that includes the positive experiences of the children and their family members, but also for the unfavorable consequences. Implementing psychosocial screening in clinical practice can facilitate the discussion of relevant topics with parents. Only inquiring the general well-being of parents will likely not suffice to uncover potential needs of families. Further, our findings imply that there should not only be attention for the well-being of mothers, but also for that of fathers and for the entire family system.

Chapter 7 provides a reflection on the main findings in the light of existing literature. Further, the clinical implications of our findings are discussed, as well as the limitations of our studies. Finally, future research directions are suggested concerning the role of T4 in early life, development in children with DS, and psychosocial outcomes in families of children with DS.
**KEY MESSAGES**

- Early T4 treatment in children with DS does not result in significant long-term developmental benefits.
- Early T4 treatment does appear to have long-term effects on growth, particularly in children with neonatal elevated TSH.
- There may more than one single profile of relative strengths and weaknesses in adaptive functioning in children with DS.
- Early developmental assessments, combined with gender and the occurrence of infantile spasms can predict the level of intellectual functioning at the age of 10 years to some extent.
- Parents’ subjective evaluation of functioning is often similar to that of parents of children without DS, but when prompted parents of children with DS may report concrete consequences.
- Important themes for parents of school age children with DS that appeared from this thesis are social functioning, partner relation, vitality, participation in activities, and emotional functioning.
- Fathers of children with DS should not be forgotten; they report no less consequences than mothers.
- A proactive approach to psychosocial guidance for families of children with DS is needed; this can be facilitated by psychosocial screening with special attention for concrete consequences.