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# Chapter 9

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## **General discussion**

The aim of this thesis is to provide an insight into the impact of Down syndrome on growth, development and social functioning.

Individuals with Down syndrome constitute a recognizable group in society. With the introduction in 2002 of prenatal screening in the Netherlands, using the first-trimester combined test, it was first believed that this population may decline and Down syndrome may become a rare condition. However, in an eleven year Dutch birth cohort (1997-2007) it was observed that the prevalence of Down syndrome has not decreased. An estimated 245 children with Down syndrome are born alive each year and Down syndrome is still the most prevalent cause of intellectual impairment. This makes it important to explore the impact of Down syndrome on fundamental aspects of health and wellbeing.

John Langdon Down not only described the external characteristics of children with Down syndrome in 1866, but also their behavior and verbal ability: 'They have considerable power of imitation, even bordering on being mimics. They are humorous and a lively sense of the ridiculous often colors their mimicry' and 'They are usually able to speak; the speech is thick and indistinct, but may be improved very greatly by a well-directed scheme of tongue gymnastics'.<sup>1</sup>

Although these basic aspects received attention from the first description of the syndrome, in research the main focus still is on the physical aspects of the syndrome and not on the social and emotional issues. Improvements in the medical care of concomitant disorders are crucial in preventing illness or even death and have increased life expectancy considerably in the past two decades. Survival at the age of 20 year was 77.5% in children with Down syndrome born in 1985-1990 and has increased to 90.7% in children with Down syndrome born in 1997-2003.<sup>2</sup> Because physical health is a basic prerequisite for daily functioning, early detection of co-morbidity is essential and needs disorder specific screening programs (such as screening for hypothyroidism) as well as growth monitoring.

### **Growth monitoring**

The new disorder specific charts we designed reflect healthy growth with respect to height, weight and head circumference in persons with Down syndrome. By using these charts, secondary growth abnormalities may be detected more accurately. The earlier problems with weight and height are detected, the greater the health benefits can be. The established growth charts demonstrate the three age periods when height differences between children with and without Down syndrome increase: during pregnancy, during the first three years of life, and during puberty. Further research is needed to provide physiological clarification on the nature of this growth retardation.

It may also be possible to achieve some health benefits with regard to weight. Alarming prevalences of overweight and obesity from a young age are observed in Dutch children

with Down syndrome. This is the case in those who are otherwise healthy as well as in those with severe congenital heart defects. Professionals, parents and perhaps individuals with Down syndrome themselves should be aware of the risks accompanying overweight and obesity. Excessive weight gain should be prevented by using appropriate interventions. Specific programs which are suitable for children with Down syndrome need therefore to be developed.

### **Level of functioning**

In contrast to the intensive research concerning physical disorders in Down syndrome, research on development, behavior and social functioning is less common. Our studies provide new insights into the level of functioning of children and adolescents with Down syndrome. A major developmental delay is generally observed. At the age of 8 years, children with Down syndrome show an average developmental delay of 4 years and at the age of 16-19-years the majority of adolescents with Down syndrome have limited abilities to perform complex tasks, leading to serious difficulties in social functioning. So, despite increased stimulation of development, increased opportunities to participate and increased acceptance in society, all individuals with Down syndrome remain quite dependent.

Even those adolescents with the best scores on level of functioning, who are able to cook a basic meal without assistance and to pay in a shop, for example, still have serious difficulties in general functioning. When adolescents and young adults with Down syndrome master essential life skills it may improve their capacity for independence and reduce the intensity of support needed. Therefore, when coaching children and adolescents with Down syndrome, professionals and parents need to focus on those essential basic skills that are likely to increase the degree of independence.

### **Consequences for society**

A consequence of the lack of sufficient evidence is that, in general, incorrect assumptions are made concerning individuals with Down syndrome. It is generally believed that children with Down syndrome are charming, friendly and joyful, without scientific evidence to support this assumption. Cebula commented on this when describing social cognition in children with Down syndrome: 'In the case of Down's syndrome, an additional factor contributing to this paucity of research may be the stereotypical perception that children with Down's syndrome are highly sociable and have good 'people' skills.<sup>1,3,4</sup> This has led to a widely held assumption that their social understanding is relatively intact'.<sup>5</sup> This impression probably holds because of their high compliance: compliant children are easily accepted in society and as a result not much attention is paid to these particular aspects. However, our results show that most adolescents and young adults with Down syndrome have serious difficulties in active social functioning and experience more behavioral problems than their peers without Down syndrome. Many adolescents have problems with social interaction,

some trouble with processing information and with understanding conversations. Examples of behavioral problems are: being too dependent on adults, having obsessive thoughts, being restless and being impulsive (dealing without thinking). Many adolescents are also withdrawn and have difficulty with change.

These social and behavioral problems of individuals with Down syndrome need to be recognized as they impede optimal daily functioning and opportunities to participate in society. In particular the high levels of social problems – measured at the age of 8 years as well as at 16-19 years – have major implications. It needs to be realized that the stereotype of Down syndrome – that they are charming, friendly and joyful – fails to acknowledge the extent of behavioral problems that limit adolescents with Down syndrome in their (social) functioning. These limited abilities will hamper participation in activities and social engagement. Results suggest that this is an area where significant overall health improvement needs to be made. Medical care for children and adults with Down syndrome should focus not only on physical health, but also on what is needed for an optimal quality of life and an improved level of functioning.

### **Recommendations for counseling**

It is important to adjust the general perceptions concerning the opportunities for children and adults with Down syndrome. This implies that (expecting) parents need to be informed and counseled concerning the extremely limited chances that their child will ever be able to live independently as an adult. Positive and negative aspects need to be balanced in the information provided for professionals and parents. Most adolescents and young adults with Down syndrome may master skills such as personal hygiene, answering a telephone or swimming. But it is also necessary to acknowledge that only one third of adolescents with Down syndrome are able to spend a few hours alone at home and/or can phone other people. This means that two thirds continuously need intensive supervision. It is important to realize that only a small minority can perform more complex tasks such as cooking, shopping and cycling in traffic. Our results indicate that most tasks can only be performed in standard and predictable situations and we found that these adolescents are generally unable to handle any changes and new or unexpected situations.

Professionals are responsible for giving appropriate information to parents and colleagues. They have to outline the full picture of the way in which individuals with Down syndrome will function. It is further essential that suitable information is available to answer parents' questions. Parents of an older child have other needs than expectant parents. Availability of appropriate information material, such as images and videos, is essential. Preferably, expectant or new parents of infants with Down syndrome should also receive information from parents of adolescents or young adults with Down syndrome who can relate their

personal experiences. In this way, they will have a better opportunity to form a correct image of the future they can expect for and with their child.

### **Recommendations for further research**

- The stability in the prevalence of Down syndrome in the Netherlands and the fact that the population of children with Down syndrome remains constant indicate that medical and social facilities for their special needs remain necessary. Therefore, research into these needs must continue.
- Prenatal screening for Down syndrome has been introduced in the Netherlands for all pregnant women in order to allow them and their partners to either prepare themselves for the birth of an infant with Down syndrome or to terminate the pregnancy if Down syndrome is diagnosed. In view of the low uptake of prenatal screening by Dutch women and the observed stable prevalence of Down syndrome, it seems that the abovementioned second choice is not widely used. The reasons behind the low uptake of prenatal screening should be explored.
- Calculations are proposed to estimate the expected final height in children with Down syndrome. The Target Height calculations were derived under the assumption that the correlation between mid-parental height standard deviation (SD) and child height SD in Dutch children with Down syndrome is identical to the general Dutch population. Further research is needed to investigate whether this assumption is justified.
- Children with Down syndrome show growth retardation during critical periods when the highest growth velocity occurs. This finding indicates the time intervals in which further growth might possibly be achieved. Therefore, future research should focus on the exact qualities of the observed deflections in growth of otherwise healthy children with Down syndrome: is their growth spurt restricted by an inborn lower growth velocity or do other reversible phenomena play a role?
- Criteria are needed to define abnormal growth. No specific criteria have been proposed for abnormal growth on the charts for children with Down syndrome. The utility of the referral criteria for the general Dutch population as presented in the guideline 'Detection and referral criteria in short stature' has not been tested for growth in children with Down syndrome. Further research is necessary to see whether such referral criteria are equally suitable for children with Down syndrome.

- More information concerning the underlying cause of excessive weight gain in children with Down syndrome is necessary for developing strategies for prevention and intervention. From the information from studies among children in the general population, we assume that physical activity and/or eating patterns are most likely to be the important factors influencing body weight in children with Down syndrome. Further research is needed to establish the merit of this assumption and to explore other possible underlying factors.
- Excessive weight gain in children and adults with Down syndrome should be prevented. We expect that a structured healthy life style (including a healthy diet and sufficient physical activity) will be especially effective in children with Down syndrome, due to their habit of keeping to a strict routine. Specific validated prevention programs suitable for children with Down syndrome to help prevent excessive weight gain and to support their families should be tested and become widely available if proved suitable.
- Children with Down syndrome experience various behavioral problems, for example they are withdrawn, are dependent on adults, their coordination is clumsy and they have obsessive thoughts. It may be possible to significantly improve their behavior, especially the social aspects. From an early age onwards, individuals with Down syndrome should be stimulated to develop better social skills. Special programs for this should be developed and evaluated.
- Factors influencing the social participation and development of children with Down syndrome, as well as the relationship between developmental level and problem behavior, and its influence on quality of life, need to be investigated.
- Recurrent respiratory tract infections in children with Down syndrome, as reported by parents, are associated with more delayed development, more behavioral problems and a lower health-related quality of life (HRQoL). Further research is needed to investigate whether the observed association is based on a causal relationship. Many variables that influence development, behavior and HRQoL, for example gender and congenital heart defects, cannot be changed. However the problem of recurrent respiratory tract infections could potentially be influenced by improved medical care and this could possibly lead to better functioning in these children.
- The level of impairment of the humoral immune system and its contribution to recurrent respiratory tract infections in Down syndrome has not been fully elucidated; more research on this topic is needed. Also, more insight into causative pathogens may lead to specific preventive interventions, i.e. prophylactic antibiotics or additional immunizations.

- Adolescents with Down syndrome in our sample have limited abilities to perform the relatively more complex tasks needed for independent living. This leads to dependency on others and to serious difficulties in social functioning. Further research is needed to develop and study the effectiveness of intervention programs to specifically improve specific social functioning skills of children with Down syndrome.
- The gap between our research findings and evidence-based interventions and effective educational approaches needs to be bridged. Furthermore, more detailed knowledge of how social cognition of children with Down syndrome develops throughout childhood and which factors contribute to better social functioning in these children need to be studied. These results may provide additional information on the most appropriate ways to support social development in children with Down syndrome at various ages.
- Although small studies show some evidence for positive short-term effects of stimulation by early intervention programs, there is no evidence that they lead to long-term benefits. Further evidence is needed to provide knowledge about what can (or cannot) be gained by stimulation programs. The limits of these programs need to be known to prevent too high expectations beforehand and disappointment afterwards. It is essential that expectations are realistic.
- At 8 years, as well as at 16-19 years of age, individuals with Down syndrome experience more emotional and behavioral problems than other children. Further research is needed to investigate if problems at late-adolescence can be predicted by the profile of behavioral problems in childhood.
- The causes of the behavioral problems are unknown. It is obvious, that these problems are caused by the genotype of Down syndrome as well as by factors influencing the phenotype. The 'dual diagnosis' of intellectual disability and psychopathology needs to be studied in adolescents with Down syndrome.
- Appropriate definitions of psychopathology in Down syndrome (such as attention deficit and hyperactivity disorders (ADHD) and autism) are needed. Special screening instruments for these problems should be developed. Sufficient information concerning the clinical implications and possible treatments of these behavioral disorders is essential for provision of optimal care.



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