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Growth, development and social functioning of individuals with Down syndrome

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Citation

Gameren-Oosterom, H. B. M. (2013, June 19). *Growth, development and social functioning of individuals with Down syndrome*. Retrieved from <https://hdl.handle.net/1887/20980>

Version: Corrected Publisher's Version

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Title: Growth, development and social functioning of individuals with Down syndrome

Issue Date: 2013-06-19

Chapter 1

General introduction

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Down syndrome

Trisomy of chromosome 21, as discovered by Lejeune in 1959, is the cause of Down syndrome.¹ Little is known about the causes of non-disjunction that lead to the trisomy. However, its consequences are well known: delayed cognitive and motor development and physical disorders. Children and adults with Down syndrome are easily recognized by their external features, which were first described by the British medical doctor John Langdon Down, to whom the syndrome owes its name.²

Medical care

Children with Down syndrome need special medical care. Guidelines for this specific care have been developed, in the Netherlands as well as in other countries.³⁻⁷ In 2011 a completely revised version of the Dutch guideline for medical care of children with Down syndrome was released by the Pediatric Association of the Netherlands.³ This guideline aims to offer directions for optimal medical care by pediatricians, who work together with other (medical) specialists, e.g. youth health care physicians, physicians for the intellectually disabled, social workers and parent organizations. Early detection and treatment of concomitant disorders, facilitating best possible cognitive and motor development and support to achieve active participation in society, are important aspects of this guideline.

One of the major focuses in the medical care for neonates with Down syndrome is on cardiology, as 43-58% of children with Down syndrome are born with a congenital heart defect.⁸ The most common are atrioventricular septum defects and ventricle septum defects. In the Netherlands, each neonate with Down syndrome receives a cardiac ultrasonography investigation within the first four weeks of life, in order to detect a congenital heart defect early. As a result of this early detection, improved surgical possibilities and clinical care, mortality in the first year of life (which is mainly influenced by congenital heart defects) has declined to 4% in the Netherlands in 2003.⁹⁻¹¹ Congenital gastrointestinal tract malformations are also frequently seen in children with Down syndrome (prevalence of 4-10%).¹²

During childhood, children with Down syndrome are at an increased risk for developing hypothyroidism. At adolescence the prevalence of hypothyroidism is 2-5%.¹³ Because of the approximately 50 times higher prevalence of acquired hypothyroidism in childhood and the signs and symptoms which overlap with the Down syndrome phenotype, it is advised to screen for hypothyroidism actively by checking thyroid function annually. A coeliac disease screening program is also in place for children with Down syndrome in the Netherlands.

Prenatal screening

In the Netherlands, in 2002 screening using the first-trimester 'combined test' was introduced in a nonsystematic manner and only at the pregnant women's request.^{14,15}

The test includes an assay of the serum concentrations of pregnancy-associated plasma protein A (PAPP-A) and the free β subunit of human chorion gonadotrophin (f β -hCG) between 9-14 weeks of pregnancy and an ultrasound measurement of nuchal translucency between 11-13⁺⁶ weeks of pregnancy. The risk of Down syndrome is calculated based on a combination of the results of these tests, maternal age and pregnancy duration. Fetal karyotyping is offered if the risk is ≥ 1 in 200.

Up to 2002, only women aged 36 years or older and those with a family history of chromosomal abnormalities were offered prenatal screening for Down syndrome, using chorion villous sampling or amniocentesis.

Growth

Optimal physical growth is a reflection of good health. In the Netherlands, physical growth is being monitored during childhood and is evaluated to signal deviation of growth and allow optimal health care. To monitor growth it is necessary to have access to appropriate and up-to-date growth charts.¹⁶ It is well known that children with Down syndrome have growth retardation and a specific growth pattern. Since growth assessment depends on the growth pattern characteristic for a specific syndrome, disorder specific charts are desirable to monitor growth.¹⁷

These specific charts need to reflect optimal growth. Therefore, in establishing growth charts, it is important to be aware that children with Down syndrome have a high risk of developing many disorders known to influence growth. Only those children with Down syndrome who are otherwise healthy can achieve optimal growth. References for optimal 'healthy' growth can allow health care professionals to monitor the growth of individual children with Down syndrome and identify relative growth retarding comorbidities at an early stage.

Development to independent social functioning

An important feature of children with Down syndrome is their intellectual impairment with delayed cognitive and motor development.¹⁸ The level of functioning determines how the child functions in everyday life and the extent of support needed. The development of children with Down syndrome will also be influenced by behavioral problems. Children with Trisomy 21 are prone to psychopathology, although the risk is lower than in children who have other syndromes causing intellectual disability or children with nonspecific intellectual disability.^{19,20}

A policy of stimulating children with Down syndrome from a young age has been introduced during the last decennia. Development of children with Down syndrome is often stimulated by using early intervention programs, rearing at home instead of in an institution and integration in mainstream schools. It is generally believed that increased stimulation and acceptance creates opportunities for people with Down syndrome to participate in society.

Various small studies on the short-term effects of early stimulation in Down syndrome have been published; some showing positive short-term effects on specific areas and some showing no effects of stimulation.²¹⁻²³ There is no evidence that these programs provide long-term benefits.²³ It is reasonable to expect that stimulation will have specific short term direct effect on what children learn, but these effects cannot be extrapolated to all areas of learning and to long-term effects. Furthermore, it is questionable if it can be realistically expected that stimulation can lead to a sufficiently increased developmental level to enable children with Down syndrome to function independently as (young) adults.

When a child with Down syndrome is born, parents want to be reliably informed about the expected development and predicted quality of life of their child. Most of the currently available information focuses on the medical aspects and physical disorders. It is extremely important that appropriate information on expected mental and social development is also available.

Knowledge gap

It is problematic that realistic and actual information on growth, development and social functioning in children, adolescents and young adults with Down syndrome is lacking.

Up to 2010, the available growth charts for Dutch children with Down syndrome were based on the growth of children who attended special schools and who were measured in 1989. In order to take the secular trend and the influence of co-morbidity on growth into account, new up-to-date growth references are needed. In addition, the increasing prevalence of overweight and obesity worldwide in children is alarming and needs attention, also in children with Down syndrome.

Development and behavioral problems in Down syndrome have been studied frequently during the past 50 years. However, the majority of these studies are not population-based, included fewer than 50 children with Down syndrome, or date back to the 1970s and 1980s, when the children grew up under different circumstances. Care for individuals with Down syndrome has improved during the past two decades and, consequently, life expectancy is considerably improved. However, the potential effect of these improvements on development, behavior and social functioning is less clear than the effect on life expectancy. Scarce any population based information is available on the actual expected development and level of social functioning and on the 'dual diagnosis' of intellectual disability and psychopathology.

Aims of the thesis

We aim (A) to evaluate trends in the prevalence of Down syndrome in the Netherlands; (B) to explore growth of children with Down syndrome and provide updated growth references for height, head circumference and weight and (C) to investigate levels of development, social

functioning and behavior in children and adolescents with Down syndrome. In this thesis we provide insight into the impact of Down syndrome on growth and development, from conception to adulthood.

These aims have been translated into the following five research questions:

1. What is the trend in the prevalence of Down syndrome in the Netherlands in the period 1997-2007?
2. What is the growth pattern of otherwise healthy children with Down syndrome in the age range of 0-18 years?
3. What are the prevalence rates of overweight and obesity in Dutch children with Down syndrome and are these influenced by concomitant disorders?
4. How is the general level of development, behavior and health-related quality of life of 8-year-old children with Down syndrome?
5. What degree of independent social functioning do adolescents with Down syndrome actually reach?

Outline of the thesis

In the first part of this thesis, **chapter 2** provides an evaluation of the trends in the prevalence of children with Down syndrome in the Netherlands, based on an eleven year birth cohort (1997-2007). In the second part of the thesis several aspects of growth in children with Down syndrome are presented. In **chapter 3** height and head circumference are described, including references for healthy Dutch children with Down syndrome and comparisons with height and head circumference references from the general population. In **chapter 4** weight is addressed, including the prevalence of overweight and obesity in children with Down syndrome.

The third part of this thesis includes a description of the level of functioning of children with Down syndrome, assessed at 8 years of age. This starts with an overview of the level of development, problem behavior and quality of life in **chapter 5**. In addition, in **chapter 6** the association between concomitant recurrent respiratory tract infections and level of development in eight-year-olds with Down syndrome is presented.

In the fourth part of this thesis the level of functioning at adolescence is being presented. For these studies, our cohort is assessed at the age of 16-19 years. In **chapter 7** the degree of independent social functioning at adolescence is described. **Chapter 8** describes the observed behavioral problems. Finally, the main findings are summarized in the fifth part. The results are discussed and implications for professional practice and further research are presented in **chapter 9**. **Chapter 10** contains a summary of the results. Table 1.1 shows all studies presented in these chapters.

Table 1.1: *Studies presented in this thesis*

	Study population	Data source	N	Focus of the study
Part 1 Introduction	Nationwide birth cohort (1997-2007)	National register	3169	- Prevalence of Down syndrome
Part 2 Growth	Dutch children aged 0-26 years	Retrospective from medical records	1596	- Growth of height and head circumference - Prevalence of overweight
Part 3 Development and behavior in childhood	Nationwide cohort of children born in 1992-1994, assessed at 8 year	Home visits and parent questionnaires	337	- Development, problem behavior and quality of life - Impact of recurrent respiratory tract infections on development
Part 4 Social functioning and behavior at adolescence	Nationwide cohort of children born in 1992-1994, assessed at 16-19 year	Parent questionnaires	322	- Social and practical skills - Problem behavior

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