

MEDICAL FACT SHEET

DSAN Objectives:

- *To advocate for people with Down Syndrome in places of Learning, Health Care and Work through all available communication channels, DSAN website, electronic and printed media coverage, outreach events, etc.*
- *To reach out to parents of people with Down Syndrome and provide practical support to them in terms of Health Care, Education and Work opportunities.*

As parents of a child with DS we may not be medical experts but we certainly have been exposed to some of the medical challenges you may or may not experience with your child with DS.

Some medical conditions are more prevalent in people with DS and a number of medical checks are recommended as part of the **Early Intervention** approach.

Information provided here is given from a parents' perspective. You will need to decide which information you find appropriate and useful in your situation, preferably in consultation with your health professional.

As mothers and fathers we advise you to carefully consider recommended medical interventions and only agree to those that are strictly necessary. We wish to caution against subjecting our children with DS to overly frequent and/or invasive medical testing and procedures.

We recommend you to talk to other parents of a child with DS, do research on the internet, speak honestly and openly with your health practitioner and work out a medical testing schedule that suits your child.

Find below a Medical Check List compiled from the following sources (*though mostly based on the Medical Guideline "Medische Leidraad" as published on the Dutch Down Syndrome Association's site (first reference below)*) and supplemented by own experience:

- Stichting Down Syndroom, The Netherlands – <http://www.downsyndroom.nl>
- About.Com – <http://downsyndrome.about.com>
- The Paediatric Orthopaedic Society of North America – <http://www.posna.org>
- Lucile Packard Children's Hospital at Stanford – <http://www.lpch.org>

- The Down's Syndrome Medical Interest Group – <http://www.dsmig.org.uk>
- Coeliac UK – <http://www.coeliac.org.uk>

We also advise you to make use of the growth charts for children with DS so you will be in a better position to monitor the growth of your child. You will find the growth charts for girls with DS [here](#) and for boys with DS [here](#).

Please contact DSAN for additional information and how to get into contact with parents/caretakers of people with DS who have indicated willingness to share their medical experiences with you.

	What is that - in simple terms:	What test is done - in simple terms:	Notes:
Paediatric Cardio Check	To check heart function and possible hole in the heart.	Sonar of the heart and possibly further testing.	If heart abnormality is suspected immediately at birth, otherwise at 3 months of age. Repeat heart check if first check was done at less than 3 months of age.
Obstipation	Obstipation is a condition where the bowel never fully empties out. Unlike constipation, obstipation develops over a matter of many months many times at least a year.	There is no specific diagnostic test for obstipation. Diagnosis is often made on the basis of the patient's symptoms and medical history. The primary symptom of obstipation is severe constipation with frequent passing of hard stool, typically every 3 to 5 days.	
Coeliac disease	Coeliac disease (pronounced see-liac, spelt celiac disease in other countries) is an autoimmune disease. Gluten, which is found in wheat, barley and rye triggers an immune reaction in people with coeliac disease. This means that eating gluten damages the lining of the small intestine. Other parts of the body may be affected.	Blood test to check for anti-endomysium, (IgA-antigliadine), (total IgA).	Routine screening at 2 years of age, earlier on indication.
Obstructive apnea syndrome	Obstructive sleep apnea occurs when a child stops breathing during periods of sleep. The cessation of breathing usually occurs because of a blockage (obstruction) in the airway. Tonsils and adenoids may grow to be large relative to the size of a child's airway (passages through the nose and mouth to the windpipe and lungs). Inflamed and infected glands may grow to be larger than normal, thus causing more blockage. Periods of blockage occur regularly throughout the night and result in a poor, interrupted sleep pattern. If this pattern continues, the lungs and heart may suffer permanent damage.	Specialized sleep test which is done at home using a small machine issued by the testing centre.	
Ear Nose Throat (ENT) Test for Hearing	This is a normal hearing test using various techniques to determine if the child hears well. Sometimes we think our child does not understand what we are saying while in fact he/she simply cannot hear us well.	Hearing test.	

<p>Atlantoaxial Instability (AAI)</p>	<p>Atlantoaxial instability (AAI) is a common orthopedic problem seen in people with Down syndrome. Although it has a complicated name and sounds somewhat intimidating, for the most part it causes no problems to those who have it. Since people with DS have low muscle tone and lax ligaments, their vertebrae can become misaligned. When the C1 and the C2 vertebrae are misaligned, you have AAI. There are two types of atlantoaxial instability - asymptomatic AAI and symptomatic AAI. Asymptomatic AAI means that AAI can be seen on an x-ray, but it is not causing any neurologic problems in the person who has it. Symptomatic AAI means that AAI is present on an x-ray, and it is causing some neurologic problems for the person that has it. Somewhere between 10% to 20% of people with DS have asymptomatic AAI on x-ray, and only 1% to 2% of people with DS have symptomatic AAI.</p>	<p>Most cases of asymptomatic AAI are made through screening x-rays. The diagnosis of symptomatic AAI is usually made either through neurological exam (physical exam that examines how the nerves are working) and/or by x-ray.</p>	
<p>Hips/Patella</p>	<p>Typically the risk of dislocation of the hips and knees is identified early during (neuro) physio therapy (see below) but may need special attention in people with DS who have a high risk (approx. 20%).</p>	<p>X-rays</p>	<p>Cervical spine dislocation is another risk for some people with DS. Participation in the Special Olympics requires cervical spine screening.</p>
<p>Thyroid function</p>	<p>If the thyroid gland is overactive (hyperthyroidism) a person may be agitated and jittery, lose weight and suffer palpitations. If the gland is underactive (hypothyroidism) a person can become tired, overweight and generally sluggish with slow physical and mental reactions. People with DS do sometimes have an overactive thyroid, but it is far more common for them to have an underactive thyroid gland. 15 – 20% of adolescents with DS the thyroid gland is not working properly. Not enough thyroxine is produced to keep the body running at an optimum rate. These people benefit greatly from thyroid replacement therapy. This involves taking one or more tablets of a thyroid preparation every day.</p>	<p>Blood test to check for free T4 + TSH, (antibodies once every 2nd year).</p>	<p>There are homeopathic, non-chemical solutions available in the market, also in Namibia.</p>
<p>Eye Test</p>	<p>Regular check up with specially trained optician or specialist eye doctor.</p>	<p>Regular eye test.</p>	
<p>Speech Therapist</p>	<p>Children with DS have difficulty with speech, typically due to low muscle tone – tongue is a muscle – and possibly also due to lesser cognitive ability.</p>	<p>Test at qualified speech therapist.</p>	<p>Extending speech beyond age of 4 is found beneficial to most children with DS.</p>
<p>Neuro/Paediatric Physiotherapy</p>	<p>To improve muscle tone and coordination, early physio therapy is highly recommended. Many children with DS are born with hypotonia, which means low muscle strength. This will help your child with rolling, crawling, sitting, standing and eventually walking and general good body coordination.</p>	<p>On-going checks during physio therapy sessions.</p>	
<p>Dental checks</p>	<p>Regular check up with dentist.</p>	<p>Regular check up with dentist.</p>	<p>Twice yearly! People with DS typically experience challenges with their teeth, also due to relatively small mouth cavity and typically regular use of anti-biotics negatively affects teeth.</p>

	Months		Years																	
	0-3	4-12	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18
Paediatric Cardio Check	0-3 m		1 y																	
Obstipation	0-3 m	4-12 m	1 y	2 y	3 y															
Coeliac disease				2 y																
Obstructive apnea syndrome	0-3 m	4-12 m	1 y	2 y	3 y	4 y	5 y	6 y												
Ear Nose Throat (ENT) Test for Hearing		4-12 m	1 y	2 y	3 y	4 y		6 y		8 y		10 y		12 y		14 y		16 y		18 y
Atlantoaxial Instability (AAI)			1 y	2 y	3 y	4 y	5 y	6 y	7 y	8 y	9 y	10 y	11 y	12 y	13 y	14 y	15 y	16 y	17 y	18 y
Hips/Patella	0-3 m	4-12 m	1 y	2 y	3 y	4 y	5 y	6 y	7 y	8 y										
Thyroid function		4-12 m	1 y	2 y	3 y	4 y	5 y	6 y	7 y	8 y	9 y	10 y	11 y	12 y	13 y	14 y	15 y	16 y	17 y	18 y
Eye Test	0-3 m		1 y		3 y		5 y		7 y		9 y		11 y		13 y		15 y		17 y	
Speech Therapist	0-3 m	4-12 m	1 y	2 y	3 y	4 y														
Neuro/Paediatric Physiotherapy	0-3 m	4-12 m	1 y	2 y	3 y	4 y	5 y	6 y	7 y	8 y										
Dental checks			1 y	2 y	3 y	4 y	5 y	6 y	7 y	8 y	9 y	10 y	11 y	12 y	13 y	14 y	15 y	16 y	17 y	18 y