

DOWN SYNDROME

A. Medical problems

Newborn

- cardiac defects (50%): AVSD [most common], VSD, ASD, TOF or PDA
- gastrointestinal (12%): duodenal atresia [commonest], tracheo-oesophageal fistula, anorectal malformation, pyloric stenosis and Hirschsprung disease.
- vision: congenital cataracts (3%), glaucoma.
- hypotonia & joint laxity
- feeding problems. Usually resolve after a few weeks.
- congenital hypothyroidism (1%)
- congenital dislocation of the hips

Infancy and Childhood

- delayed developmental milestones
- mild to moderate intellectual impairment (IQ 25 to 50)
- seizure disorder (6%)
- recurrent respiratory infections
- hearing loss (>60%) due to secretory otitis media, sensorineural deafness, or both
- visual impairment – squint (50%), cataract (3%), nystagmus (35%), glaucoma, refractive errors (70%)
- sleep related upper airway obstruction. Often multifactorial.
- leukaemia (relative risk:15 to 20 times). Incidence 1%
- atlantoaxial instability. Symptoms of spinal cord compression include neck pain, change in gait, unusual posturing of the head and neck (torticollis), loss of upper body strength, abnormal neurological reflexes, and change in bowel/bladder functioning. (see below)
- hypothyroidism (10%). Prevalence increases with age
- short stature – congenital heart disease, sleep related upper airway obstruction, coeliac disease, nutritional inadequacy due to feeding problems and thyroid hormone deficiency may contribute to this
- over/underweight

Adolescence and Adulthood

- puberty - in *Girls* menarche is only slightly delayed. Fertility presumed - in *Boys* are usually infertile due to low testosterone levels
- increased risk of dementia /Alzheimer disease in adult life
- shorter life expectancy

Management

- communicating the diagnosis is preferably handled in private by a senior medical officer or specialist who is familiar with the natural history, genetic aspect and management of Down syndrome.
- careful examination to look for associated complications.
- investigations: 1. echocardiogram by 2 weeks (if clinical examination or ECG were abnormal) or 6 weeks. 2. Chromosomal analysis. 3. T4 /TSH at birth or by 1-2 weeks of life.

Table 1. Incidence of Down syndrome

Overall Incidence: 1 in 800-1000 newborns	
Maternal Age-Specific Risk for Trisomy 21 at livebirth	
Age (years)	Incidence
20	1 in 1500
30	1 in 900
35	1 in 350
40	1 in 100
41	1 in 70
42	1 in 55
43	1 in 40
44	1 in 30
45	1 in 25

Source Hecht and Hook '94

- early intervention programme should begin at diagnosis if health conditions permit
- assess strength & needs of family. Contact with local parent support group should be provided (Refer list of websites below)
- health surveillance & monitoring: see Table 5

Atlantoaxial instability

- seen in X rays in 14% of patients; symptomatic in 1-2%.
- small risk for major neurological damage but cervical spine X rays in children have no predictive validity for subsequent acute dislocation/ subluxation at the atlantoaxial joint
- children with Down's syndrome should not be barred from taking part in sporting activities
- appropriate care of the neck while under general anaesthesia or after road traffic accident is advisable

Table 2. Karyotyping in Down syndrome

Non-disjunction trisomy 21	95%
Robertsonian Translocation	3%
Mosaicism	2%

Recurrence Risk by Karyotype

Nondisjunction Trisomy	
47(XX or XY) + 21	1%
Translocation	
both parents normal	low; <1%
other carrier	10%
father carrier	2.5%
either parent t(21q;21q)	100%
Mosaics	< 1%

Useful websites

- The Down Syndrome Medical Interest Group (UK): www.dsmig.org.uk
- Down Syndrome: Health Issues: www.ds-health.com
- Growth charts for children with Down Syndrome: www.growthcharts.com
- Educational issues: www.downsed.org
- Kiwanis Down Syndrome Foundation: www.kdsf.netmyne.com
- Educational & support centre. <http://www.disabilitymalaysia.com/>
- Parents support group. <http://groups.yahoo.com/group/DownSyndromeMalaysia>
- Jabatan Pendidikan Khas. <http://www.moe.gov.my/jpkhas/>.
- Jabatan Kebajikan Malaysia. <http://www.jkm.gov.my/>

Table 5. Recommendations for Medical Surveillance for children with Down Syndrome

	Birth - 6 weeks	6 - 10 months	12 months	18 mths - 2½ yrs	3 - 3½ years	4 - 4½ years
Thyroid blood tests ¹	T4 & TSH		T4 & TSH including antibodies		T4 & TSH including antibodies	
Growth monitoring ²	Length, weight and head circumference checked regularly and plotted on Down's syndrome growth charts.	Visual behaviour: Check for congenital cataract	Visual behaviour: Check for congenital cataract	Orthoptic examination, refraction and ophthalmic examination ³	Visual acuity, refraction and ophthalmic examination ³	Length and weight should be checked at least annually and plotted on Down's syndrome growth charts.
Eye check	Visual behaviour: Check for congenital cataract	Visual behaviour: Check for congenital cataract	Orthoptic examination, refraction and ophthalmic examination ³	Visual acuity, refraction and ophthalmic examination ³	Visual acuity, refraction and ophthalmic examination ³	Visual acuity, refraction and ophthalmic examination ³
Hearing check	Neonatal screening, if locally available	Full audiological review (hearing, impedance, otoscopy) by 6-10 months and then annually				
Heart check and other advice	Echocardiogram 0-6 weeks			dentist advice		
Age 5 to 19 years						
Paediatric review	Annually					
Hearing	2 yearly audiological review (as above)					
Vision / Orthoptic check	2 yearly					
Thyroid blood tests	At age 5 years, then 2 yearly					
School performance	Check performance and placement					
Sexuality & employment	To discuss when appropriate during adolescence					
<p>Note: The above table are suggested ages. Check at any other time if there are parental or other concerns. Perform developmental assessment during each visit.</p> <p>(adapted from Down Syndrome Medical Interest Group (DSMIG) guidelines)</p>						
<p>footnote:</p> <ol style="list-style-type: none"> 1. Asymptomatic patient with mildly raised TSH (not greater than 10mU/l) but normal T4 does not usually warrant treatment but should be tested more frequently because of increased likelihood of developing uncompensated hypothyroidism. 2. Down syndrome centile charts available (www.growthcharts.com). Consider using weight for length chart of typically developing children for weight assessment. Those with BMI > 98th centile or underweight should be referred for nutritional assessment & guidance. Thyroid function should be checked if there's accelerated weight gain. 3. Performed by optometrist/ophthalmologist 						