Medical Management of Children & Adolescents with Down Syndrome in Ireland

APPROVED GUIDELINES

Down’s Syndrome Medical Interest Group (DSMIG) (UK & Ireland)

Department of Paediatrics
University of Dublin, Trinity College
The National Children’s Hospital, AMNCH, Tallaght
Medical Management of Children & Adolescents with Down Syndrome in Ireland

Approved Guidelines

Professor Hilary MCV Hoey    Joan Murphy RSCN MSc Paediatrics
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INTRODUCTION

Down Syndrome is the most common congenital cause of developmental disability in Ireland with a birth prevalence of 1 in 546 live births, which is the highest in Europe. It is well recognised that as a group they have a high incidence of treatable medical disorders. All studies show that early intervention carries a better outcome for their general health, quality of life and life expectancy. With medical progress many now live into their sixties.

In order to assess the medical and psychosocial needs of children and adolescents with Down syndrome in Ireland we conducted a surveillance study in the Eastern Health Board (now the Eastern Regional Health Authority).

Medical guidelines were originally developed by the Down Syndrome Medical Interest Group for use in the United Kingdom. Our research provided the necessary evidence based data to construct medical management guidelines appropriate for children and adolescents with Down syndrome in Ireland. Many health care professionals with expertise in the management of children and adolescents with Down syndrome in Ireland have contributed to the Irish guidelines. These guidelines have now been approved by The Irish College of General Practitioners, The Faculty of Paediatrics of The Royal College of Physicians of Ireland and the Faculty of Public Health Medicine of The Royal College of Physicians of Ireland. Implementation of these guidelines is now urgently required.

We are very grateful to Dr. Jennifer Dennis, Director of Information and Research, DSMIG, Dr. Liz Marder, Vice Chairman DSMIG and all members of the group for the enormous amount of work that they have undertaken in the development of the guidelines.

We are also very grateful to the many Irish health professionals who supported and assisted us in carrying out the study and in particular we wish to express our sincere thanks to all the parents and children who took part in the study.

We wish to thank Dr. Sheila Macken, Mr. Don McShane, Dr. Philip Mayne, Dr. Desmond Duff, Dr. Myra O’Regan, Ms Aoiife Walsh, Mr. Michael O’Keefe, Mr. Esmond Fogarty, the late Dr. Zachary Johnson, Ms. Virginia Delaney, Dr. Siobhan Murnaghan, Dr. Frances Kelly, Professor Denis Gill, Dr. Owen Hensey, Dr. Mary McKay, Dr. Edwina Daly, Dr. Edna Roche, Dr. Judith Meehan, Ms Mary Cronin, Mr. Michael Harney, Dr. Colm Costigan, Dr. Louis Ramsey, Dr. Jervais Corbett, Bro. Finnian Gallagher, Dr. Brendan McCormick, Dr. Noel McDonnell, Dr. Mary Staines, Dr. Mona Byrne, Dr. Martin McLauglin, Dr. Mona O’Donnell, Professor O. Conor Ward, Dr Austin O’Carroll, the Area Medical Officers in the Eastern Regional Health Authority and all the Paediatricians and Staff in the three Children’s Hospitals and the Developmental and Educational Centres, who supported us in many ways throughout the study and accommodated us at all times.

We are very grateful to Footsteps, Down Syndrome Ireland, the Minister for Health and Children and the Provost, University of Dublin, Trinity College for sponsoring this Research. We also thank the National Children’s Hospital Foundation for sponsoring the scientific meeting to launch the guidelines in May 2001.

Professor Hilary MCV Hoey
BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME

GROWTH

Short stature is a recognised characteristic of most people with Down syndrome. Average height of children with Down syndrome at most ages is around the 2nd centile for the general population. For the majority the cause of growth retardation is not known. Some conditions leading to poor growth - congenital heart disease, sleep related upper airway obstruction, coeliac disease, nutritional inadequacy due to feeding problems, and thyroid hormone deficiency occur more frequently among those with the syndrome. Regular surveillance of growth, general health, nutritional and thyroid status should aid in early identification of pathological causes of growth retardation.

UK/ Ireland growth charts for healthy children with Down syndrome from birth to 18 years are now available. These reference values are essential for assessing linear growth. However, as many older children and adults with the syndrome tend to be overweight, the reference values for weight should not be used as a standard that children should aim to achieve. Instead the body mass index (BMI) data included on the charts should be used to aid the assessment of overweight.

Guidelines:
1. We suggest that it is good practice to record and chart height and weight frequently in the first two years using Down syndrome specific charts. Thereafter measurements should be made at least annually throughout childhood and at regular intervals in adult life. Regular measurements of this sort are likely to be sensitive early indicators of the many medical problems which are over represented in the syndrome.

2. Children normally loose weight after birth but regain the weight by approximately day 10. Preliminary data suggest that many babies with Down syndrome do not regain birth weight until around 1 month of age. This is not reflected in the growth charts because of their cross sectional nature. This early failure to thrive is usually due to feeding difficulties many of which resolve after the first few weeks. From 1 month weight should increase parallel to the centiles. Failure to do so should be investigated.

3. Of those with measurements below the 2nd centile some will have major pathology but some may be failing to thrive for other reasons – e.g. feeding difficulties. Such children should have their dietary intake evaluated and may need to be referred to a paediatrician or paediatric endocrinologist for assessment.

4. The Down syndrome specific growth charts clearly reflect the tendency to excess weight gain among the UK and Irish study sample particularly in later childhood. Hence standard BMI charts have been included on the growth charts. We suggest that all those over age 5 years with weight above the 75th centile should be charted on these BMI charts. Those above the 91st BMI centile should be carefully monitored. Those above the 98th BMI centile should be considered for further assessment and guidance.

5. Although there is a high prevalence of overweight/obesity among people with Down syndrome this is not inevitable. As with the general population weight is influenced by environmental as well as biological factors.
6. Appropriate anticipatory guidance regarding diet and physical activity should be given for all those with the syndrome.

7. Thyroid function should always be checked in those with accelerated weight gain.

8. In childhood growth spurts and plateaux occur as in all children but among the Down syndrome population these tend to be more prolonged. They are not reflected in the smoothed curves of a standardised chart.

9. The Down syndrome specific chart suggests an absence of pubertal growth spurt. However those with the syndrome do have an adolescent growth spurt. It is usually less vigorous than in the general population. Puberty may occur at an earlier age and requires anticipation together with education and support for parents and child. Early onset of puberty has a limiting effect on final height.

10. As with all children, head circumference should be measured regularly and charted on Down syndrome specific charts. If there is any cause for concern subsequent measurements should be made.

11. The use of growth hormone in Down syndrome is still being evaluated. There is no evidence that it should be prescribed except in the unusual situation of concurrent primary growth hormone deficiency.

12. The influence of parental height on target height appears to be variable.

UK/Ireland Down’s Syndrome Specific Growth Charts are available from Harlow Printing Ltd, South Shields, Tyne and Wear. NE33 4PU. Tel 0044 191 455 4286

References (Growth)


Dr Jennifer Dennis      Director of Information and Research DSMIG (UK & Ireland)

Professor Hilary MCV Hoey      Joan Murphy RSCN MSc Paediatrics
BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME

CARDIAC DISEASE
CONGENITAL AND ACQUIRED

1. Between 40 and 50% of babies with Down syndrome have congenital heart defects. Of these 30-40% have complete atrioventricular septal defects (AVSD).\(^1,2\) Most AVSD can be successfully treated if the diagnosis is made and the baby referred for full corrective surgery before irreversible pulmonary vascular disease is established.\(^3\)

2. There must be a high level of clinical suspicion of congenital heart disease for all newborns with the syndrome

3. It is essential to establish the cardiac status of every child by age 6 weeks.\(^4-10\)

4. Clinical examination alone is insufficient to detect even *some of* the most serious abnormalities.\(^2,8,11\)

5. It is very unlikely that a serious abnormality requiring early intervention (AVSD) will be missed if one of the following courses of action is taken.\(^2,10,12\)
   
   **(a)** Clinical examination plus electrocardiogram (ECG) and chest X-ray (CXR) for all newborns and again at age 6 weeks, followed by echocardiography only for those with abnormal findings.
   
   **or**
   
   **(b)** Clinical examination, ECG plus echocardiogram in the newborn period, both carried out by an appropriate person (see below 6).

   However, even if early investigations are reported as 'normal', if a child develops signs or symptoms of cardiac disease appropriate investigations must take place, as structural problems may not have been evident at an earlier age.

6. It is not always essential to refer newborn babies with the syndrome to a cardiologist. However, all clinical examinations should be by a doctor experienced in the care of newborns; an experienced paediatrician should review CXR and ECG findings; echocardiograms should be carried out and reviewed by staff with appropriate paediatric experience *under the supervision of a paediatric cardiologist*. *Telemedicine may provide a useful intermediate step between paediatrician and cardiologist.* Those with suspected problems should be referred for immediate cardiological review so that intervention, if necessary, can take place before pulmonary vascular disease develops.

7. It is recognised that minor heart defects (atrial septal defect and small ventricular septal defects) may be missed in those children who do not have echocardiograms but these should declare themselves clinically, as for any child, in the normal course of child health surveillance.

8. Parents and carers of all children with Down syndrome with heart lesions should be given verbal and written information about infective carditis preventive measures.
9. It should be remembered that despite a normal echo at birth children with Down syndrome, like all other children, can develop symptoms and signs of heart disease at a later age e.g. secondary to airway problems.\textsuperscript{13}

10. There is an increased incidence of mitral valve prolapse and of aortic regurgitation in adults. This has implications for infective carditis prevention particularly because of the high incidence of periodontal disease among this population. We therefore recommend an echo screen for all people with Down's syndrome early in adult life.\textsuperscript{9,14}

11. If a potential risk situation for infective endocarditis arises for an adult with Down syndrome who has not had an adult echo, preventive prophylactic measures should be started.

References (Heart disease)


Dr Jennifer Dennis      Director of Information and Research DSMIG (UK & Ireland)

We are very grateful to Dr Desmond Duff, Consultant Cardiologist, Our Lady’s Hospital for Sick Children, Crumlin for his support with the development of the guidelines for children and adolescents with Down syndrome in Ireland

Professor Hilary MCV Hoey       Joan Murphy RSCN MSc Paediatrics
BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME

THYROID DISORDER

1. At all ages thyroid disorder (usually hypothyroidism) occurs more frequently in people with Down syndrome than in the general population. Around 10% of the school age population have uncompensated hypothyroidism. The prevalence increases with age. If undiagnosed, thyroid disorder constitutes a significant cause of preventable secondary handicap. Diagnosis on clinical grounds is unreliable. Biochemical screening is essential. As in the general population those with significant abnormalities of any thyroid function test (TFT) should either be treated (if there is uncompensated hypothyroidism) or kept under close clinical and biochemical surveillance.

2. All babies in the U.K and Ireland have a neonatal screen for hypothyroidism.

3. Biochemical testing, including estimation of T4, TSH, and thyroid antibodies should be carried out at least once every two years from age 1 year and throughout life.

4. Information is currently coming in from several areas where the feasibility of fingerprick TSH Guthrie screening is being investigated. Preliminary evaluation suggests that this may prove an effective screening procedure, which may be possible annually, once the appropriate structures, personnel and funding are in place.

5. Transient changes may occur. Mildly raised TSH (5-10mU/l) or the presence of antibodies with normal T4 and no clinical evidence of hypothyroidism may not warrant treatment. It does however indicate increased likelihood of developing uncompensated hypothyroidism. Such people should therefore be tested more frequently than those with normal test results. A specialist opinion may be required.

6. Clinicians should always bear in mind the high prevalence of thyroid disorder in people with Down syndrome and have a low threshold for testing thyroid function if there is any clinical suspicion at times between biochemical testing.

7. As in the general population key clinical pointers are lethargy and/or changes in affect, cognition, growth, or weight.

8. Consideration of hypothyroidism is mandatory in the differential diagnosis of depression and dementia.

9. The possibility of hyperthyroidism should also be born in mind.

References (Thyroid disorder)


Dr Jennifer Dennis      Director of Information and Research DSMIG (UK & Ireland)

We are very grateful to Dr. Philip Mayne, Consultant Chemical Pathologist, The Children’s University Hospital, Temple Street, for his support in the development of the guidelines for children and adolescents with Down syndrome in Ireland.

Professor Hilary MCV Hoey      Joan Murphy RSCN MSc Paediatrics
1. There is a high prevalence of ocular disorder among people with Down syndrome. Refractive errors and strabismus (squint) may occur at an early age and persist into childhood\(^1,2,3,4,5\). Over 54\% of children with Down syndrome will require glasses in primary school\(^1\). The majority of children with Down syndrome have reduced accommodation at near (this means that they do not focus accurately on near targets),\(^2,6,7\). Cataract and/or Glaucoma may occur in infancy\(^8\). Cataract extraction in our population of children with Down Syndrome is a safe and effective procedure with a very encouraging visual outcome\(^9\). Nystagmus is present in 18\%\(^10\) and Brushfield Spots are present in the eye in many children at birth. Keratoconus\(^11\) and cataract may develop in adolescents and young adults\(^12\). Untreated disorders which cause vision problems are a significant cause of preventable secondary handicap and require increased observation at all ages\(^5\).

2. All newborns with Down syndrome should have an eye examination carried out at 4-6 weeks to exclude congenital glaucoma, cataract and other eye abnormalities\(^13\) and thereafter should be included in community screening programmes.

3. Visual behaviour must be monitored by a paediatrician before their first formal ophthalmologic review. Those who start to squint or show other abnormalities of gaze, visual behaviour or attention should be referred for ophthalmological review.

4. Between 18 months and 2 years all children with Down syndrome should have a formal ophthalmological examination. This should include orthoptic assessment, refraction and fundus examination. At least one third will have ocular/visual defects by this age\(^1,14\). Those with deviation from normal should be kept under appropriate specialist review.

5. Refractive errors, most commonly hypermetropia (long-sightedness), which often reduce spontaneously in other children, are likely to persist beyond infancy in children with Down syndrome\(^6,2\). Correction for hypermetropia may be helpful at a younger age than that for typically developing children especially since the majority will have defective accommodation\(^15\)\(^2,6,7\). Distance and near functioning visual acuity and accommodative ability should be checked at every review and a prescription for near correction or bifocals considered for all children of school going age\(^16,2\).

6. A further formal ophthalmological examination should be performed at around 4 years of age\(^1,17,13\). At this age at least 50\% are likely to have refractive errors\(^1\).

7. After the age of 4 years vision and refractive error should be checked at least every 2 years throughout life by professionals (optometrists or ophthalmologists & orthoptists) with appropriate skills and expertise in managing this client group\(^18\). If hypermetropia is not present at age 4 years it is not likely to occur later on, but myopia may develop at any age\(^5,2\).

8. Children and adults with Down syndrome should be expected to respond to standard vision testing procedures at appropriate developmental age but a distraction free environment and extra time may be necessary to optimise performance. Others may
require more specialized visual tests. Distance and near functioning visual acuity and accommodation (focussing ability) should be checked at every review. Detail vision (i.e. visual acuity) is likely to remain poorer than expected throughout life even when appropriate spectacles are worn.19,20

9. Blepharitis, (inflammation of the eyelids with redness at the edge of the lids and crusting around the lashes) has been reported to occur in up to 30% of children with Down syndrome10,21 and can be managed in the usual way.22 Nasolacrimal duct obstruction also occurs commonly21,23 and may need specialist referral

10. Local optometrists give an excellent service but subjects who are difficult to examine in this setting should be referred to a specialist clinic.

11. As with all children, if at any age visual acuity deteriorates a specialist opinion is required.

12. Any child or adult with pain, and/or changing vision, visual disturbance and/or red eye, should be referred for urgent specialist opinion.

References (Ophthalmic problems)


**Reviewed and Updated July 2009 with guidance and expertise from Dr. Mary Cregg, Optometrist and Professor Michael O'Keeffe, Consultant Ophthalmologist.**

Dr Jennifer Dennis Director of Information and Research DSMIG (UK & Ireland)

We are very grateful to Professor Michael O’Keefe, Consultant Ophthalmologist, The Children’s University Hospital, Temple Street, for his support with the development of the guidelines for children and adolescents with Down syndrome in Ireland. May 2003

Professor Hilary MCV Hoey Dr Joan Murphy RCN, MSc PhD
1. Well over 50% of people with Down syndrome have significant hearing impairment, which may be mild, moderate, severe or profound (30 - >95 dB HL). Sensorineural and/or conductive loss may be present at any age. Hearing impairment can be successfully managed in this population. If undetected it is likely to be a significant cause of preventable secondary handicap. Lifelong audiological surveillance is essential for all. The main cause of conductive loss is persistent otitis media with effusion (OME, glue ear). The natural history of OME and response to intervention differ from that in the general population hence local surveillance and management protocols need to be set up specific to people with Down’s syndrome.

2. People with Down syndrome of all ages should have rapid access to specialist audiology services.

3. Because of an increased incidence of congenital sensorineural loss newborns with Down syndrome should be included in neonatal screening programmes where available. This does not preclude the need for ongoing surveillance.

4. Guidance for parents of children with Down syndrome should include discussion about hearing problems and their management, supported by good quality written information.

5. Whether or not a baby with Down syndrome has passed a neonatal screen all should have full audiological assessment between age 6 and 10 months. This should include measurement of auditory thresholds, impedance testing and otoscopy. To ensure inclusion of the child with Down’s syndrome participation in existing child health hearing surveillance programmes should be encouraged.

6. Therefore by 10 months it should have been established whether or not a child with Down syndrome has any degree of permanent hearing loss with or without OME. A clear management plan must have been agreed with the parents and intervention instigated where necessary.

7. In the second year (usually around 18 months) all children with Down syndrome – whatever their previous hearing status - should have further audiological review carried out in a manner appropriate for a child with learning disabilities. This should include assessment of auditory thresholds, impedance testing and otoscopy. This should be repeated at least yearly until age 5 and thereafter 2 yearly for life. More frequent testing will be necessary if problems exist.

8. Transition of care from paediatric to adult services should involve direct transfer of care to a named person.

9. At all ages people with Down's syndrome have narrow ear canals, which predispose to accumulation of wax. This may affect impedance testing and hearing. Early management to clear wax would be desirable to remove any further impact on hearing loss.
10. Most people with Down syndrome are able to respond to standard tests – e.g. distraction; speech discrimination; pure tone audiometry (play or standard); and visual reinforcement audiometry. These tests must be performed by professionals trained in audiology with experience in working with people with learning disabilities. Threshold measurement tests appropriate to developmental age must be used.\(^5\)\(^,\)\(^{21}\)

11. Because of increased incidence of sensorineural as well as conductive loss the frequency range tested should include 8000Hz whenever feasible as failure at this level may be an early warning of impending high frequency sensorineural deafness.\(^3\)\(^,\)\(^{23}\)

12. Diagnostic Auditory Brain Stem (ABR) responses in people with Down syndrome must be interpreted with caution.\(^2\)\(^,\)\(^{23}\) *A Child with Down syndrome with a failed ABR may require Oto Acoustic Emissions (OAEs) to distinguish cochlear from neurological pathology.*\(^6\)

13. As in the general population all those who are hearing impaired should have access to specialist hearing support services (Speech and Language Therapy; Teachers of the deaf; etc)

14. At all ages particular attention should be paid to the treatment of suppurative nasal and ear conditions.\(^4\)\(^,\)\(^{20}\)

15. In adults with the syndrome hearing assessment is essential in the differential diagnosis of depression and dementia.\(^3\)

**References (Hearing impairment)**


Dr Jennifer Dennis  Director of Information and Research DSMIG (UK & Ireland)

We are very grateful to Mr. Don McShane, Consultant ENT Surgeon, Mr. Michael Harney, Senior ENT Registrar and Ms Aoife Walsh, Senior Clinical Audiologist, The National Children’s Hospital, AMNCH for their support with the development of the guidelines for children and adolescents with Down syndrome in Ireland.

Professor Hilary MCV Hoey  Joan Murphy RSCN MSc Paediatrics
BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME.

CERVICAL SPINE INSTABILITY

1. People with Down syndrome have a small risk for acute or chronic neurological problems caused by cervical spine instability.1,2

2. Currently there is no screening procedure, which can predict those at risk. In particular cervical spine x-rays in children have no predictive validity for subsequent acute dislocation/subluxation at the atlantoaxial joint.3-7

3. Children with Down syndrome should not be barred from sporting activities because there is no evidence that participation in sports increases the risk of cervical spine injury any more than for the general population.6,8

4. Although the risk of injury is small, if any child or adult with Down syndrome needs an anaesthetic, the anaesthetist and recovery room staff must always be reminded of the diagnosis, so that appropriate care can be taken to avoid cervical injury, whilst manipulating the head and neck in the unconscious subject.9

5. Although the risk of injury is small, if a person with Down syndrome is involved in a road traffic accident personnel involved in their care should be alerted to the possibility of cervical spine instability and of the need for particular care relative to this.1,5

6. If a person with Down syndrome develops pain behind the ear or elsewhere in the neck, abnormal head posture, torticollis, deterioration of gait, manipulative skills, or bowel and/or bladder control they should be referred immediately to an appropriate specialist (usually a neurologist or a spinal orthopaedic surgeon).

References (Cervical spine instability)


Dr Jennifer Dennis      Director of Information and Research DSMIG (UK & Ireland)

We are very grateful to Mr. Esmond Fogarty, Consultant Orthopaedic Surgeon, The National Children’s Hospital, AMINCH, and Our Lady’s Hospital for Sick Children, Crumlin for his support with the development of the guidelines for children and adolescents with Down syndrome in Ireland.

Professor Hilary MCV Hoey      Joan Murphy RSCN MSc Paediatrics
# DOWN SYNDROME MEDICAL MANAGEMENT GUIDELINES

*Suggested schedule of health checks taken from Guidelines*

<table>
<thead>
<tr>
<th>Age</th>
<th>Growth assessment as above at each routine visit*</th>
<th>Heart</th>
<th>Thyroid</th>
<th>Sight</th>
<th>Hearing</th>
</tr>
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<tbody>
<tr>
<td>Birth to 6 wks</td>
<td>Length/weight/head circumference – Plot on Down Syndrome Specific Growth Charts*</td>
<td>Clinical Examination Echocardiogram 0-6 weeks or Clinical Examination ECG + Chest X-ray Birth and 6 wks</td>
<td>Routine Guthrie test</td>
<td>Eye Examination, check for congenital cataract, congenital glaucoma + any other eye abnormality</td>
<td>Neonatal screening where available</td>
</tr>
<tr>
<td>6-10 months</td>
<td>Growth assessment as above at each routine visit*</td>
<td>Dental Advice</td>
<td>Full Thyroid function tests or TSH (finger prick)** yearly when available</td>
<td>Visual behaviour, check for squint</td>
<td>Full audiological review (Otoscopy, Impedance, Hearing thresholds)</td>
</tr>
<tr>
<td>12 months</td>
<td>Growth assessment as above at each routine visit*</td>
<td>Dental Advice</td>
<td>Full Thyroid function tests or TSH (finger prick)** yearly when available</td>
<td>Visual behaviour, check for squint</td>
<td></td>
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<tr>
<td>18-24 months</td>
<td>Growth (height/weight) assessment as above*</td>
<td>Dental Advice and Examination of teeth</td>
<td>Full Thyroid function tests or TSH (finger prick)** yearly when available</td>
<td>Ophthalmological examination including Orthoptic screening, refraction and fundal examination and focusing ability</td>
<td>Full audiological review as above</td>
</tr>
<tr>
<td>3 – 3½ years</td>
<td>Growth (height/weight) assessment as above*</td>
<td>Dental Advice and Examination of teeth</td>
<td>Full Thyroid function tests or TSH (finger prick)** yearly when available</td>
<td></td>
<td>Full audiological review as above</td>
</tr>
<tr>
<td>4 – 4½ years</td>
<td>Growth (height/weight) assessment as above*</td>
<td>Dental Advice and Examination of teeth</td>
<td>Full Thyroid function tests or TSH (finger prick)** yearly when available</td>
<td>Ophthalmological examination as above</td>
<td>Full audiological review as above</td>
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*Encourage a healthy lifestyle (healthy eating and regular exercise) at all times

**TSH(finger prick)- capillary whole blood thyroid stimulating hormone (TSH) sample – using one circle on National Newborn Screening Programme card

*From age 5years to 19 years*

Paediatric Medical Review Annually

<table>
<thead>
<tr>
<th>Medical Area</th>
<th>Details</th>
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<tr>
<td>Cardiology</td>
<td>Echo in early adult life to rule out mitral valve prolapse</td>
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<tr>
<td>Hearing</td>
<td>2 yearly audiological review as above</td>
</tr>
<tr>
<td>Vision</td>
<td>2 yearly Ophthalmological examination including refraction and fundal examination</td>
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<tr>
<td>Thyroid</td>
<td>2 yearly from 5 years (venous) or TSH (fingerprick)** annually, when appropriate structures, personnel and funding are in place</td>
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Updated 30th September 2009
**IRISH HEALTH CARE PROFESSIONALS**
Who supported the study and development of the guidelines

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<td>Consultant Paediatrician, St. Michael’s House Clinic, Goatstown, Dublin 14</td>
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<tr>
<td>Professor Denis Gill</td>
<td>Consultant Paediatrician and Paediatric Nephrologist, The Children’s Hospital, Temple Street, Dublin 1</td>
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<td>Dr. Owen Hensey</td>
<td>Consultant Paediatrician, The Children’s Hospital, Temple St./CRC Clontarf</td>
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<tr>
<td>Dr. Mary McKay</td>
<td>Consultant Paediatrician/Paediatric Accident &amp; Emergency Medicine, The National Children’s Hospital, AMINCH, Tallaght, D 24</td>
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<tr>
<td>Dr. Edwina Daly</td>
<td>Consultant Paediatrician, The National Children’s Hospital, AMINCH, D 24</td>
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<tr>
<td>Ms Mary Cronin</td>
<td>Manager, St. Catherine’s Centre, Newcastle, Co. Wicklow</td>
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<tr>
<td>Mr. Michael Harney</td>
<td>Senior ENT Registrar, The National Children’s Hospital, AMINCH, Tallaght, D 24</td>
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<tr>
<td>Dr. Colm Costigan</td>
<td>Consultant Paediatrician/Endocrinologist, Our Lady’s Hospital for Sick Children, D 12</td>
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<tr>
<td>Dr. Louis Ramsay</td>
<td>Consultant Psychiatrist, Medical Director, St. John of God Brothers, Stillorgan, Kildare Glenageary Road and Celbridge Co Kildare</td>
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<tr>
<td>Dr Jervais Corbett</td>
<td>Consultant Paediatrician, Stewart’s Hospital for the Mentally Handicapped, Palmerstown, Co. Dublin</td>
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<tr>
<td>Bro. Finnian Gallagher</td>
<td>Director, St. John of God Brothers, Menni Services, Island Bridge, Dublin 8</td>
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<tr>
<td>Dr. Brendan McCormick</td>
<td>Consultant Psychiatrist, Cheeverstown House, Mental Handicap Centre, Kilveroe, Templeogue, Dublin 6W</td>
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<tr>
<td>Dr Noel McDonnell</td>
<td>Consultant Psychiatrist, Medical Director, St. Michael’s House Dublin</td>
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<tr>
<td>Dr. Mary Staines</td>
<td>Consultant Psychiatrist, Medical Director, Steward’s Hospital for the Mentally Handicapped, Palmerstown, Co. Dublin</td>
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<tr>
<td>Dr Mona O’Donnell</td>
<td>Area Medical Officer, SWAHB, Newbridge Health Centre, Newbridge, Co. Kildare</td>
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<tr>
<td>Dr Mona Byrne</td>
<td>Paediatrician, Cheeverstown House, Mental Handicap Centre, Kilveroe, Templeogue, Dublin 6W</td>
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<tr>
<td>Dr. Martin McLauglin</td>
<td>Medical Director, St. Vincent’s Centre, Navan Road, Dublin 7.</td>
</tr>
<tr>
<td>Area Medical Officers</td>
<td>All AMOs in 10 Health Areas in the Eastern Regional Health Authority</td>
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</tbody>
</table>
## LIST OF CONTACT ADDRESSES

<table>
<thead>
<tr>
<th>Organization</th>
<th>Address</th>
<th>Phone</th>
<th>Fax</th>
<th>Website</th>
<th>Email</th>
</tr>
</thead>
<tbody>
<tr>
<td>Department of Paediatrics, Trinity College and The National Children’s Hospital, AMINCH, Tallaght, Dublin 24</td>
<td><a href="http://www.amnch.ie">www.amnch.ie</a></td>
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<td>Fax 01 4626593/863786</td>
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<tr>
<td>Down Syndrome Ireland (Head Office)</td>
<td>Citylink Business Park, Old Naas Rd., Dublin 12</td>
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<tr>
<td>DSMIG/DSMIS</td>
<td>The Children’s Centre, City Hospital Campus, Hucknall Road, Nottingham, NG5 1PB</td>
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<tr>
<td>Cheeverstown House, Mental Handicap Centre, Kilvere, Templeogue, D6W</td>
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<td>Early Support Team, Sisters of Charity of Jesus &amp; Mary, Moore Abbey, Monasterevan, Co. Kildare</td>
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<tr>
<td>Kare, Lr. Eyre Street, Newbridge, Co. Kildare (Parent and friends org.)</td>
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<td>Our Lady's Hospital for Sick Children, Crumlin, Dublin 12.</td>
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<tr>
<td>St. Catherine’s Centre, Newcastle, Co. Wicklow</td>
<td>Early Services</td>
<td>01-2819485</td>
<td>01-2812392</td>
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<tr>
<td>St. John of God Brothers, Kildarten (Dunmore House) 111 Upr. Glenageary Road, Co. Dublin.</td>
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<td>St. John of God Brothers, Menni Services, Islandbridge, Dublin 8.</td>
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<tr>
<td>St. John of God Brothers, St. Raphael’s, Celbridge, Co. Kildare.</td>
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<tr>
<td>St. John of God Brothers, Stillorgan Road, Stillorgan, Co. Dublin.</td>
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<tr>
<td>St. Michael’s House, Developmental Clinic, Ballymun Road, Dublin 7.</td>
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<tr>
<td>St. Michael’s House, Grosvenor Road, Rathgar, Dublin 6.</td>
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<td>St. Vincent’s Centre, Daughters of Charity, Navan Road, Dublin 7.</td>
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<tr>
<td>Stewart’s Hospital for the Mentally Handicapped, Palmerstown, Co. Dub</td>
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<tr>
<td>The Children’s Hospital, Temple Street, Dublin 1</td>
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</tbody>
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Down’s Syndrome Medical Interest Group (DSMIG UK & Ireland)
Down’s Syndrome Medical Information Services (DSMIS)

DSMIG was launched in 1996. It is a network of doctors from the UK and Ireland whose aim is to share and disseminate information about the medical aspects of Down’s syndrome and to promote interest in the specialist management of the syndrome. The group meets twice a year at the Royal Society of Medicine in London.

Initiatives to date include:

- The production of guidelines for basic medical surveillance essentials for people with Down’s syndrome
- The production and nationwide distribution of a special insert for babies born with Down’s syndrome for the UK national parent held personal child health record (PCHR)
- Production of UK/Ireland growth charts for children and adolescents with Down’s syndrome.
- Organisation of regional DSMIG road shows for health care professionals
- Setting up of Down’s Syndrome Medical Information Services (DSMIS) – the information arm of DSMIG The remit of this organisation is to provide information to the health care professionals about the medical aspects of Down’s syndrome. This service is complementary to that provided for parents by the National Down’s Syndrome Support Groups
- Launch of a temporary website in 2000 with a projected launch date for a full information site in June 2001

We have no health service funding or corporate sponsorship. We have to date received financial help towards specific projects and administration costs from the DSA: Marks and Spencer plc; Mencap City Foundation; The David Solomon Trust; Harlow Printing; and Children Nationwide. We ourselves accrue some monies from fees charged for medical road shows; from occasional personal donation of lecture fees etc and from Royalty payments on the growth charts and PCHR

Currently we work under the charitable umbrella of the Nottingham Community Health NHS Trust Charitable Funds but are seeking charitable status in our own right.

DR JENNIFER DENNIS (DSMIG UK & Ireland)
Director of Information and Research

DSMIG, The Children’s Centre, City Hospital Campus, Hucknall Road, Nottingham, NG5 1PB
Tel: 0044 115 962 7658 (ext: 45667) Answer phone: 0044 115 934 5502
Fax 0044 115 962 7915 Email: info@dsmig.org.
BASIC MEDICAL SURVEILLANCE
ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME

Guidelines of the
Down’s Syndrome Medical Interest Group
DSMIG (UK & Ireland)
IRISH EDITION

We are grateful to the Association for the Prevention of Disabilities for financial support for initial meetings of the surveillance essentials development group and to the Learning Disability Forum of the Royal Society of Medicine and Mencap City Foundation for ongoing support for a number of initiatives.
DOWN’S SYNDROME SURVEILLANCE GUIDELINES

Background notes
(As service delivery varies in different countries some minor adjustments to the guidelines have been made for Ireland and these appear in italics)

People with Down syndrome do not have unique medical problems, which differ from the general population. However some medical conditions are heavily over represented among those with the syndrome. Most of these are treatable disorders, which if undiagnosed, impose an additional but preventable burden of secondary handicap.

These surveillance guidelines have been developed on the basis of available evidence by a group of clinicians with a special interest in Down syndrome. They are updated as new research and audit evidence becomes available. The overall aim is to help ensure equitable provision of basic essential medical surveillance for all children with Down’s syndrome in the UK and in the Republic of Ireland. The Royal College of Paediatrics and Child Health has been supportive of the venture and we have had guidance from the Centre for Evidence Based Child Health.

A set of background notes is being developed which cover the evidence on which the guidelines are based. Currently these are completed for the cardiac and hearing sections and are available electronically via jendennis@dsmig.org.uk

The guidelines are not a blueprint for Gold Star services. Their purpose is to set out a minimum safe standard of basic medical surveillance which we consider essential for all those with the syndrome. This we consider to be the identification of cardiac disease, thyroid disorder, hearing impairment and ophthalmic problems and the appropriate monitoring of growth. We have also included information which we hope will increase understanding of the complex issues surrounding cervical spine instability. We are currently producing clinical awareness notes covering other conditions which are over represented in the syndrome. In parallel with the guidelines we have produced UK/Ireland Down’s syndrome specific growth charts and a special insert for UK Personal Child Health Record (The Red Book) for children born with Down’s syndrome.

These are available from:
Harlow Printing Ltd, South Shields, Tyne and Wear. NE33 4PU. Tel 0044 191 455 4286
The guidelines are available in electronic format on www.dsmig.org.uk The site also features the PCHR insert and an order form for the growth charts and PCHR

All other enquiries to:
Down’s Syndrome Medical Information Services
Children’s Centre. City Hospital Campus. Nottingham NG5 1PB.
Tel: 0044 115 962 7658 Ext.45667. 0044 115 934 5502 (answer phone). Fax :0044 115 962 7915
Email ; info@dsmig.org.uk

Jennifer Dennis. DSMIG.
Director of Information and Research 06.06.01
Members of DSMIG Guideline Development Core Group

Chairman


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Past Medical Advisor, Irish Down’s Syndrome Association