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
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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.

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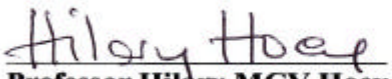

Professor Hilary MCV Hoey

Joan Murphy RSCN MSc Paediatrics, Dip Stats

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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.[1] Some conditions leading to poor growth - congenital heart disease,[2,3] sleep related upper airway obstruction,[4] coeliac disease,[5,6] nutritional inadequacy due to feeding problems, and thyroid hormone deficiency[7,8] 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.[9] These reference values are essential for assessing linear growth. However, as many older children and adults with the syndrome tend to be overweight,[10,11] 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.[12] 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.[13] 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[10,11] among people with Down syndrome this is not inevitable. As with the general population weight is influenced by environmental[11,14] 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*.[16] 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.[17-20]

12. The influence of parental height on target height appears to be variable.[21]

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

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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.[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.[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.

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

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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.[1-5] Around 10% of the school age population have uncompensated hypothyroidism. The prevalence increases with age.[6] If undiagnosed, thyroid disorder constitutes a significant cause of preventable secondary handicap. Diagnosis on clinical grounds is unreliable.[7,8] 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.[9]

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.[6,10]

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.[10,11] Mildly raised TSH *(5-10mU/l)* or the presence of antibodies with normal T4 and no clinical evidence of hypothyroidism may not warrant treatment.[12,13] 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.[14,15]

9. The possibility of hyperthyroidism should also be born in mind.[5,16]

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

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Professor Hilary MCV Hoey Joan Murphy RSCN MSc Paediatrics

BASIC MEDICAL SURVEILLANCE ESSENTIALS
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OPHTHALMIC PROBLEMS

1. There is a high incidence of ocular disorder among people with Down syndrome.[1-6] Refractive errors and Strabismus (squint) may occur at an early age. *Over 50% of people with Down's syndrome wear glasses.* Cataract and/or Glaucoma may occur in infancy.[7] Nystagmus is present in about 10% and *Brushfield Spots are present in the eye in many children at birth.* Keratoconus,[8,9] is more common in adolescents and young adults. *In particular as the patient gets older, high myopia (near sightedness), can occur.*[3] If untreated, many of these disorders are a significant cause of preventable secondary handicap.

2. All newborns with Down syndrome should have an eye examination carried out at birth to exclude congenital cataract *and glaucoma* and should thereafter be included in community screening programmes.

3. Visual behaviour must be monitored by a paediatrician. Those who start to squint or show other abnormalities of gaze, visual behaviour or attention or changes in the appearance of the eye or excessive watering in the first year of life should be referred for ophthalmological review in the normal way.

4. All children with Down syndrome should have a formal ophthalmological examination including orthoptic assessment, refraction and fundal examination during the second year of life. The majority may have some deviation from normal and should be kept under close review

5. A further *formal ophthalmological* examination should be performed at around the age of four years. If hypermetropia is not present at this age it is not likely to occur later on.

6. Children and adults with Down's syndrome *will respond to a variety of visual testing and this depends very much on their developmental age. Some will be able to perform the routine, ordinary visual testing. Others will require more specialized visual tests. A distraction free environment will optimise performance.* Distance vision should be checked at every review.

7. After the age of 4 years vision should be checked every two years *by an ophthalmologist or optometrist throughout life.*

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

9. If at age 4 years or thereafter, a child has visual acuity assessed as at least 6/9 on a linear Snellen chart then subsequent testing by the school visual clinic service is adequate.

10. Any child identified by the school visual clinic service as less than 6/9 vision should be referred to *a Community Ophthalmologist.*

11. Any child identified *with less than 6/12* on a linear Snellen chart should be referred to Community Ophthalmologist for further assessment.

12. *Local opticians* give an excellent service but subjects who are not cooperative in this setting should be referred to a specialist clinic.

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

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

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Professor Hilary MCV Hoey Joan Murphy RSCN MSc Paediatrics

BASIC MEDICAL SURVEILLANCE ESSENTIALS
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HEARING IMPAIRMENT

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).*[1-3] Sensorineural and/or conductive loss may be present at any age.[3-10] Hearing impairment can be successfully managed in this population.[11-15] If undetected it is likely to be a significant cause of preventable secondary handicap.[3,6,12,14] Lifelong audiological surveillance is essential for all.[16-21] 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.[4,6,13,15,22]

2. People with Down syndrome of all ages should have rapid access to specialist audiology services.[4]

3. Because of an increased incidence of congenital sensorineural loss newborns with Down syndrome should be included in neonatal screening programmes where available.[1,14,19] This does not preclude the need for ongoing surveillance.[16]

4. Guidance for parents of children with Down syndrome should include discussion about hearing problems and their management, supported by good quality written information.[19]

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.[10,19,21] 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.[5] 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.[7,21]

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]

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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).

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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

	Growth	Heart	Thyroid	Sight	Hearing
Birth 6 wks	Length/weight/head circumference – Plot on Down Syndrome Specific Growth Charts*	Clinical Examination Echocardiogram 0-6 weeks **or** Clinical Examination ECG + Chest X-ray Birth **and** 6 wks	Routine Guthrie test	Eye Examination, check for congenital cataract and glaucoma.	Neonatal screening where available
6-10 months	Growth assessment as above at each routine visit*			Visual behaviour, check for squint	Full audiological review (Otoscopy, Impedance, Hearing thresholds)
12 months	Growth assessment as above at each routine visit*	Dental Advice	Full Thyroid function tests **or** TSH (finger prick)** yearly when available	Visual behaviour, check for squint	
18-24 months	Growth (height/weight) assessment as above*	Dental Advice and Examination of teeth	Full Thyroid function tests **or** TSH (finger prick)** yearly when available	Ophthalmological examination including Orthoptic screening, refraction and fundal examination	Full audiological review as above
3 – 3 ½ years	Growth (height/weight) assessment as above*	Dental Advice and Examination of teeth	Full Thyroid function tests **or** TSH (finger prick)** yearly when available		Full audiological review as above
4 – 4 ½ years	Growth (height/weight) assessment as above*	Dental Advice and Examination of teeth	Full Thyroid function tests **or** TSH (finger prick)** yearly when available	Ophthalmological examination as above	Full audiological review as above

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

Cardiology Echo in early adult life to rule out mitral valve prolapse

Hearing 2 yearly audiological review as above

Vision 2 yearly Ophthalmological examination including refraction and fundal examination

Thyroid 2 yearly from 5 years (venous) **or** TSH (fingerprick)** annually, when appropriate

structures,

personnel and funding are in place

Professor Hilary MCV Hoey Dr Joan Murphy RCN MSc PhD Paediatrics

Department of Paediatrics, University of Dublin, Trinity College, at The National Children's Hospital, AMNCH, Tallaght, Dublin 24

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IRISH HEALTH CARE PROFESSIONALS
Who supported the study and development of the guidelines

Professor Hilary Hoey	Head Dept Paediatrics TCD, Consultant Paediatrician/ Endocrinologist, The National Children's Hospital, AMINCH, Tallaght, Dublin and Director of Research, Down Syndrome Ireland
Ms Joan Murphy	Research Nurse, RSCN MSc Paediatrics, Department of Paediatrics and The National Children's Hospital, AMINCH, Tallaght, Dublin
Dr. Magued Philip	Consultant Paediatrician, Bons Secours Hospital, Tralee, Co. Kerry
Dr. Sheila Macken	Consultant Paediatrician, The Children's Hospital, Temple Street and St Michael's House Services
Mr. Don McShane	Consultant ENT Surgeon, The National Children's Hospital, AMINCH, Tallaght, Dublin 24
Dr. Philip Mayne	Consultant Chemical Pathologist, The Children's Hospital Temple Street, D 1.
Dr. Desmond Duff	Consultant Cardiologist, Our Lady's Hospital for Sick Children, Crumlin, D 12.
Dr. Myra O'Regan	Senior Statistician, University of Dublin, Trinity, Dublin 2.
Ms Aoife Walsh	Senior Clinical Audiologist, The National Children's Hospital, AMINCH, Tallaght, Dublin 24
Mr. Michael O'Keefe	Consultant Ophthalmologist, The Children's Hospital, Temple Street, Dublin
Mr. Esmond Fogarty	Consultant Orthopaedic Surgeon, Adelaide & Meath Hospital incorporating the National Children's Hospital, Tallaght, Dublin 24
Ms Virginia Delaney	Research Nurse, Health Information Unit, EHRA, Dr. Steven's Hospital D 8
Dr. Siobhan Murnaghan	Consultant Paediatrician, St. Michael's House Clinic, Ballymun Road, Dublin
Dr. Francis Kelly	Consultant Paediatrician, St. Michael's House Clinic, Goatstown, Dublin 14
Professor Denis Gill	Consultant Paediatrician and Paediatric Nephrologist, The Children's Hospital, Temple Street, Dublin 1
Dr. Owen Hensey	Consultant Paediatrician, The Children's Hospital, Temple St./CRC Clontarf
Dr. Mary McKay	Consultant Paediatrician/Paediatric Accident & Emergency Medicine, The National Children's Hospital, AMINCH, Tallaght, D 24
Dr. Edwina Daly	Consultant Paediatrician, The National Children's Hospital, AMINCH, D 24
Ms Mary Cronin	Manager, St. Catherine's Centre, Newcastle, Co. Wicklow
Mr. Michael Harney	Senior ENT Registrar,The National Children's Hospital, AMINCH, Tallaght, D 24
Dr. Colm Costigan	Consultant Paediatrician/Endocrinologist, Our Lady's Hospital for Sick Children, D 1
Dr. Louis Ramsay	Consultant Psychiatrist, Medical Director, St. John of God Brothers, Stillorgan, Kildarten Glenageary Road and Celbridge Co Kildare
Dr Jervais Corbett	Consultant Paediatrician, Stewart's Hospital for the Mentally Handicapped, Palmerstown, Co. Dublin
Bro. Finnian Gallagher	Director, St. John of God Brothers, Menni Services, Island Bridge, Dublin 8
Dr. Brendan McCormick	Consultant Psychiatrist, Cheeverstown House, Mental Handicap Centre, Kilvere, Templeogue, Dublin 6W
Dr Noel McDonnell	Consultant Psychiatrist, Medical Director, St. Michael's House Dublin
Dr. Mary Staines	Consultant Psychiatrist, Medical Director, Stewart's Hospital for the Mentally Handicapped, Palmerstown, Co. Dublin
Dr Mona O'Donnell	Area Medical Officer, SWAHB, Newbridge Health Centre, Newbridge, Co. Kildare
Dr Mona Byrne	Paediatrician, Cheeverstown House, Mental Handicap Centre, Kilvere, Templeogue, Dublin 6W
Dr. Martin McLauglin	Medical Director, St. Vincent's Centre, Navan Road, Dublin 7.
Area Medical Officers	All AMOs in 10 Health Areas in the Eastern Regional Health Authority

LIST OF CONTACT ADDRESSES

Department of Paediatrics, Trinity College and The National Children's Hospital, AMINCH, Tallaght, Dublin 24 www.amnch.ie	**01 4142000 01 8963785 Fax 01 4626593/863786 Email: jpmurphy@tcd.ie**
Down Syndrome Ireland (Head Office) Citylink Business Park, Old Naas Rd., Dublin 12	**1 890 374 374 01 4266500 Fax 01 4266501 http://www.downsyndrome.ie Email: info@downsyndrome.ie**
DSMIG/ DSMIS 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. www.dsmig.org.uk**
Cheeverstown House, Mental Handicap Centre,Kilvere,Templeogue,D6W	**01-4904681/ Fax 4905753 Email: info@cheeverstown.ie**
Early Support Team, Sisters of Charity of Jesus & Mary, Moore Abbey, Monasterevan, Co. Kildare	**045-525327/ Clinic 045 529455 Fax 045 529029 Email: scjmclinic@eircom.net**
Kare, Lr.Eyre Street, Newbridge, Co. Kildare (Parent and friends org.)	**045-431544 Email: kare@kareld.com**
Our Lady's Hospital for Sick Children, Crumlin, Dublin 12.	**01 4096100 www.olhsc.ie**
St. Catherine's Centre, Newcastle, Co. Wicklow Early Services	**01-2819485 01-2812392**
St. John of God Brothers, Kildarten (Dunmore House) 111 Upr. Glenageary Road, Co. Dublin.	**01-2852900**
St. John of God Brothers, Menni Services, Islandbridge, Dublin 8.	**01-6774022**
St. John of God Brothers, St. Raphael's, Celbridge, Co. Kildare.	**01-6288161**
St. John of God Brothers, Stillorgan Road, Stillorgan, Co. Dublin.	**01-2881781**
St. Michael's House, Developmental Clinic, Ballymun Road, Dublin 7.	**01-8840200**
St. Michael's House, Grosvenor Road, Rathgar, Dublin 6.	**01-4963678**
St. Michael's House, Willowfield Park, Goatstown, Dublin 14.	**1-2987033**
St. Vincent's Centre, Daughters of Charity, Navan Road, Dublin 7.	**01-8383234**
Stewart's Hospital for the Mentally Handicapped, Palmerstown, Co.Dub	**01-6264444 www.stewartshospital.com**
The Children's Hospital, Temple Street, Dublin 1	**01 8748763 www.childrenshospital.ie**

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

Dr Jennifer Dennis. Paediatrician. Oxford.
Chairman

Dr Nick Archer. Paediatric Cardiologist. Oxford.

Dr Robert Barclay. Paediatrician. Lanarkshire.
Medical Adviser, Scottish Down's Syndrome Association

Dr Barbara Crofts. Ophthalmologist. Oxford.

Dr Bethan Davies. Audiological Physician. London.

Dr Josephine Hammond. Paediatrician. St George's. London.

Professor Hilary Hoey. Paediatrician/ Endocrinologist. Dublin.
Director of Research, Down's Syndrome Ireland

Dr Patricia Jackson. Paediatrician. Edinburgh.
Medical Adviser, Scottish Down's Syndrome Association

Professor Sam Lingam. Paediatrician. Haringey Healthcare NHS Trust.
Executive Medical Director. The Association for the Prevention of Disabilities

Dr Liz Marder. Paediatrician. Nottingham.
Medical Adviser. Down's Syndrome Association.

Dr Marion McGowan. Paediatrician.
St Helier NHS Trust. Surrey

Professor Ben Sacks. Psychiatrist in Learning Disability
Charing Cross & Westminster Medical School. London.

Mr Ross Scrivener. Research and Audit Co-ordinator
Royal College of Paediatrics and Child Health

Dr Ajay Sharma. Paediatrician.
Optimum Health Services. Southwark.

Dr Barbara Stewart. Paediatrician. Oxford.

Professor O. Conor Ward. Paediatrician/Cardiologist. Dublin
Past Medical Advisor, Irish Down's Syndrome Association