Medical Management
of
Children & Adolescents
with
Down Syndrome in Ireland

APPROVED GUIDELINES 2005
With
Updates 2009 & 2015

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

Department of Paediatrics
University of Dublin, Trinity College
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Updates July 2009 & Dec 2015

Approved Guidelines

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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 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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These guidelines have been reviewed to incorporate new evidence and where required updated in 2015. These revisions have been approved by the Clinical Advisory Group (CAG), Faculty of Paediatrics, Royal College of Physicians of Ireland in February 2016. We are particularly grateful to Dr Paul Oslizlok, Mr Esmond Fogarty, Ms Patricia Gaule and Ms Fiona McGrane for their expertise and assistance with the revision of these guidelines.

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BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME

GROWTH

(Revised December 2015)

Short stature is a recognised characteristic of most people with Down syndrome.[1, 2] 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,[3-5] sleep related upper airway obstruction[6], coeliac disease,[7-9] nutritional inadequacy due to feeding problems[10] and thyroid hormone deficiency[9,11,12] 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/Republic of Ireland growth charts for healthy children with Down syndrome from birth to 18 years are available and were revised in 2011.[13,14] These reference values are essential for assessing linear growth. However, as many older children and adults with Down syndrome tend to be overweight,[15,16] the reference values for weight may over-estimate the appropriate weight for the child and should not be used as a standard that children should aim to achieve. Instead, clinical observation and the body mass index (BMI) data included on the charts should be used to aid the assessment of those who are overweight and for the prevention of obesity.

Guidelines:

1. We suggest that it is good practice to record and chart accurate height and weight frequently in the first two years using the 2011 revised Down syndrome specific charts.[13] Thereafter, measurements should be recorded at least annually throughout childhood and at regular intervals in adult life. Regular measurements are likely to be sensitive early indicators of the many medical problems which are over-represented in this syndrome.

2. Children normally lose weight after birth but usually regain the weight lost by approximately day ten of life. Preliminary data suggest many babies with Down syndrome do not regain birth weight until around one month of age.[17] This is not reflected in the growth charts because of their cross-sectional nature and thus regular assessment is necessary from birth. This early failure to thrive is usually due to feeding difficulties, many of which resolve after the first few weeks. From one month, weight should increase parallel to the centiles. Failure to do so should be investigated. Breastfeeding should be encouraged and supported.[18,19] Preterm babies with DS (born before 37 weeks of gestation) have weights that are[20] similar to the general population and so the neonatal infant and close monitoring (NIC**A**M) chart should be used until term[21] and thereafter the DS charts should be used with measurements corrected for gestational age for at least a year.

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.[10] Such children should have their dietary intake evaluated and may need to be referred to a paediatrician or paediatric endocrinologist for assessment.[17]

4. The Down syndrome specific growth charts based on data from around 6000 measurements of 1100 children clearly reflect the tendency to excess weight gain among the children within the UK and Irish study sample particularly in later

childhood.[13,14] Hence clinical assessment together with use of the standard BMI charts, which are included on the growth charts, is required. We suggest that all those over age 2 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 encouraged to lose weight and further assessment and guidance should be considered.

5. Although there is a high prevalence of overweight/obesity among people with Down syndrome this is not inevitable.[15,16] As with the general population weight is influenced by environmental[16, 22] as well as biological factors.[23]

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 may fluctuate as in all children, but among children with Down syndrome these fluctuations may be more prolonged. They are not reflected in the smoothed curves of the current standardised growth chart.

9. The Down syndrome specific chart suggests an absence of pubertal growth spurt. Those with the syndrome do have an adolescent growth spurt, but it is usually less marked than in the general population. Puberty may occur at an earlier age and requires anticipation together with education and support for both parents and the child.[24,25] 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.[26-30]

12. The influence of parental height on target height appears to be variable.[31]

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BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME

CARDIAC DISEASE - CONGENITAL AND ACQUIRED

(Revised December 2015)

1. Between 40-60% of babies with Down syndrome have congenital heart defects.[1-3] Of these, 30-40% have complete atrioventricular septal defects (AVSD).[2-5] Most AVSDs can be successfully treated if the diagnosis is made early and the baby referred for full corrective surgery before irreversible pulmonary vascular disease (PVD) is established.[6-14]

2. There must be a high level of clinical suspicion of congenital heart disease for all newborns with the syndrome.[8,15] Those diagnosed in the early neonatal period should be referred for urgent cardiac assessment as outlined below. Babies diagnosed in the later neonatal period should have accelerated referral to a paediatric cardiologist or a paediatrician with appropriate paediatric cardiological training especially if the 6 week deadline has been exceeded.

3. It is highly desirable to establish the cardiac status of every child with Down syndrome by age 6 weeks.[8,13-15] Irreversible PVD is more likely to develop quickly in children with Down syndrome.[2,9,10,13,16] Ideally surgery for AVSD is desirable by 6 months of age.[9,13,17] Early cardiac surgery is desirable for children with Down syndrome with surgically correctable cardiac lesions to optimise outcomes.[5-12,17]

4. Clinical examination alone is insufficient to detect even some of the most serious abnormalities.[4.8,15,18]

5. It is very unlikely that a serious abnormality requiring early intervention (e.g. AVSD) will be missed if the following course of action is taken.[4,8,15,19]

   - Clinical examination, electrocardiogram (ECG) and echocardiogram (ECHO) performed by someone with appropriate paediatric cardiological training[5,8] for all newborns with Down syndrome ideally by the age of 6 weeks (in particular those with a superior QRS axis on ECG).[20]
   - Telemedicine, if available, may provide a useful intermediate step between paediatrician and cardiologist.
   - Those with suspected problems should be referred for immediate paediatric cardiological review so that intervention, if necessary, can take place before pulmonary vascular disease develops.

   Fetal Echocardiography
   Given the absence of a uniform standard of fetal echocardiography we suggest that those who have had a fetal ECHO should still follow the above neonatal pathway.
   Older children who have never had an echocardiogram
   - Those with a normal ECG, no symptoms and no abnormal clinical signs should be referred routinely for additional assessment by a clinician with appropriate paediatric cardiological training.
   - Those with symptoms and/or abnormal clinical signs or ECG abnormalities should be referred urgently for assessment

6. People with Down syndrome with heart lesions are at increased risk of infective endocarditis.[21 to 24] Therefore, parents and carers of all children with Down syndrome with heart lesions should be given verbal and written information about infective endocarditis preventive measures.[25-27]

7. 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/respiratory problems,[13, 28, 29] and may be at increased risk of developing pulmonary vascular disease and right heart failure.

8. Echocardiography may occasionally fail to diagnose AVSD and other major cardiac lesions, particularly in the first few days after birth, even when undertaken by skilled practitioners.[8] As a result, if symptoms or signs of cardiac disease are detected at any age even where the early ECHO has been reported as normal, there should be a low threshold for repeating the ECHO examination.

9. There is an increased incidence of mitral valve prolapse (MVP) and aortic regurgitation (AR) from late adolescence into adulthood in people with Down syndrome which may be asymptomatic.[22, 23,30-33] This has implications for infective endocarditis prevention particularly because of the high incidence of periodontal disease among this population. [21--27] . We therefore recommend careful cardiac evaluation including echocardiography for all people with Down syndrome early in adult life.[8,30, 33, 34]

10. MVP occasionally progresses to mitral valve regurgitation (MVR). We recommend monitoring for signs of atrial fibrillation and/or left ventricular failure[30, 31] in these patients, and some may be advised regarding restriction of competitive sporting activities.[31,3]

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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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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.

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BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME

OPHTHALMIC PROBLEMS

(Revised July 2009 Ireland)

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,[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[3,2].

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 syndrome [10,21] and can be managed in the usual way [22]. Nasolacrimal duct obstruction also occurs commonly[21,23] 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.

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BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME

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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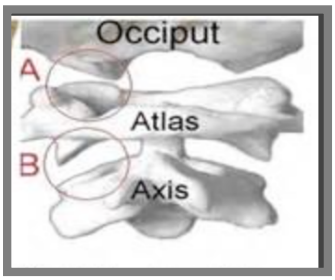
BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN SYNDROME.

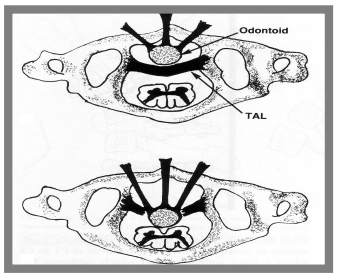
Cervical Spine Disorders: Craniovertebral Instability

(Revised and Updated December 2015)

1. People with Down syndrome have a small risk for acute or chronic neurological problems caused by cervical spine/craniovertebral instability.[1-7]



(A) Occipital-Atlantal Joint
(B) Atlantoaxial Joint



Odontoid process stable / unstable

(© Picture with kind permission of Daria Briquet 05/08/2015)

2. Currently there are no screening procedures 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[3-7] or occipito-atlanto joint.[8-14]

3. Children with Down syndrome should not be excluded 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.[7,12-17] In addition, children with Down syndrome should not be automatically excluded from participating in specialised sports (e.g. gymnastics). The requirements and clinical screening protocols of the relevant national governing bodies should be applied (www.british-gymnastics.org-Atlanto-Axial Information Pack).[18]

4. *Prior to general anaesthesia a careful history and examination should be undertaken looking for the Red Flag warning signs below. Routine pre-operative radiography is not recommended if there are no clinical concerns.*[19-21] If any child or adult with Down syndrome needs a general 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, although the risk of injury is small.[4, 9]

5. *Road Traffic Accident:* 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 craniovertebral instability and of the need for particular care relative to this.[2, 8] If there are any Red Flag warning signs detected, the patient should be immediately referred for expert opinion.

6. If a person with Down syndrome develops any of the Red Flags/Warning Signs below: 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 [4,9,22] (usually a neurologist or a spinal orthopaedic surgeon) to avoid late diagnosis of Cervical Spine Instability (CSI) with potentially devastating consequences.

Red Flags/Warning Signs:

- Neck pain, or pain behind the ear
- Abnormal head posture
- Torticollis (Wry neck)
- Reduced active neck movements
- Deterioration of gait and/or frequent falls
- Increasing fatigability on walking
- Deterioration of manipulative skills
- Loss of bowel and/or bladder function

If a person with Down Syndrome develops any of these warning signs or symptoms, ***passive movements of the neck should not be undertaken.***

7. **If a good alternative explanation for the patient's symptoms is considered likely then a careful and complete clinical history, physical and neurological examination should be undertaken <u>provided the patient has full active neck movements</u> so that they can look up at the ceiling and down at the floor, then good quality flexion and extension cervical spine x-rays should be taken. *If either clinical or radiological abnormality is found* they should be *referred immediately* to an appropriate specialist.**

8. It is essential that parents, relatives, carers and all healthcare professionals are made aware of these clinical/ Red Flags/Warning Signs and symptoms.

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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 2011 revised Down syndrome specific charts. (Use NICAM charts for preterm babies)**	**Clinical Examination** **ECG and Echocardiogram 0-6 weeks**	Routine Guthrie test	**Eye Examination; Check for congenital cataract, congenital glaucoma + any other eye abnormality**	National Neonatal Hearing screening
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, Infective endocarditis advice/information if necessary**	Full Thyroid function tests **or** TSH (finger prick)** yearly where available	**Visual behaviour, check for squint**	
18-24 months	**Growth assessment - As above*** **Chart those ≥2years of age on BMI conversion charts if concerns about overweight.**	**Dental Advice and Examination of teeth Infective endocarditis advice/information if necessary**	Full Thyroid function tests **or** TSH (finger prick)** yearly when available	**Ophthalmological examination including Orthoptic screening, refraction and fundal examination and focusing ability**	Full audiological review as above
3 – 3 ½ years	**Growth (Height/Weight) assessment and advice*. Chart on BMI conversion charts if concerns about overweight.**	**Dental Advice and Examination of teeth Infective endocarditis advice/information if necessary**	Full Thyroid function tests **or** TSH (finger prick)** yearly when available		Full audiological review as above
4 – 4 ½ years	**Growth (Height/Weight) assessment and advice as above***	**Dental Advice and Examination of teeth Infective endocarditis advice/information if necessary**	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.
Infective endocarditis information to be given later in life for those with cardiac history.

Hearing 2 yearly audiological review as above

Vision 2 yearly Ophthalmological examination including refraction and fundal examination and focusing ability

Thyroid 2 yearly from 5 years (venous) **or** TSH (fingerprick)** annually, when appropriate structures, personnel and funding are in place

A comprehensive history and careful clinical examination should be undertaken to detect other emergent health issues such as, respiratory and rheumatological complications

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Dr Mona O'Donnell	Area Medical Officer, SWAHB, Newbridge Health Centre, Newbridge, Co. Kildare
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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

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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
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