

## ***DSMIG SURVEILLANCE GUIDELINES - HEARING IMPAIRMENT BACKGROUND EVIDENCE***

People with Down's syndrome present a major challenge to general and audiological medical services as regards both the recognition and management of hearing impairment.

Estimates of prevalence of hearing impairment among this population differ according to differences in study sample attributes and assessment methods. However all studies give figures in the 50 - 100% range. For the majority the impairment is significant and requires attention<sup>3,4,5,6,23,25,37</sup>. As in the general population treatments include aiding, surgical drainage for eustachian tube dysfunction and medication<sup>27</sup>. Further evaluation of best treatment options is needed.

There is almost no scientific evidence about the effectiveness of screening populations with Down's syndrome on which to base our recommendations. One therefore has to look for evidence among the general population but this must be interpreted with caution. Not only does the prevalence, nature and natural history of the hearing impairment differ from that found in the general population (see below), in addition many of these children and adults also present challenges as far as assessment is concerned. A further confounding aspect is that, by contrast with typically developing children, clinical experience suggests that parents may have more difficulty recognising hearing problems in their Down's offspring, particularly at a young age, because of difficulty differentiating the effects from those of the syndrome itself.

### **Functional implications**

In terms of day to day living there is no reason to suggest that people with Down's syndrome are any less disadvantaged by hearing impairment than the general population.

With regard to early sensorineural loss Yoshinaga- Itano et al<sup>38</sup> have shown that in the general population diagnosis and intervention before age 6 months is essential if subsequent language deficit is to be avoided.

With regard to long term effects of significant preschool loss. In the general population there is evidence that this has adverse cognitive and behavioural implications at least until age 10<sup>2,13,10</sup>. When we searched for evidence specific to the Down's population we found only one informative paper. Whiteman et al<sup>14</sup> report in a numerically small study that only 18% of adolescents with Down's syndrome with a reported early history of otitis media had language scores above the 50th centile for the group, as against 64% of those with no such history and all of those who had been treated with tympanostomy tubes. There are many problems with this paper which are acknowledged in the text but it is currently the only evidence we have. Now that full investigation and treatment of hearing impairment in preschool children with Down's syndrome is well established we await appropriate outcome studies on populations so treated.

With regard to concurrent hearing loss. Roizen et al<sup>36</sup> were unable to demonstrate any correlation between hearing loss and early language development in a group of children with Down's syndrome age 2 - 42 months. However as the majority were less than one year old and children with Down's syndrome have in any case specific speech and language delay this finding is not surprising. Libb et al<sup>12</sup> in a study limited by small numbers and inclusion of only a few subjects with significant hearing loss nevertheless report poorer performance on intelligence tests by those school age children and young adults who have abnormal tympanograms, though no correlations with hearing threshold. Marcell<sup>31,32</sup> in a series of experiments about auditory processing difficulties in a group of adolescents with the syndrome showed correlations between hearing dysfunction and some specific language subtests. For the

adult population Keiser et al <sup>11</sup> found a moderate relationship between hearing acuity and receptive vocabulary scores. Evenhuis <sup>6</sup> emphasises the importance of assessment of hearing abilities in the differential diagnosis of dementia.

There is thus ample evidence that hearing impairment is grossly overrepresented among people with Down's syndrome. Remediation is essential in terms of day to day living. Such evidence as there is suggests that, as in the general population, treatment in the preschool years is likely to be advantageous in the long term. However there are practical problems as to how most efficiently to identify these impairments at a population level at different ages.

## Screening issues

### The first 6 months

There is anecdotal evidence of increased congenital sensorineural loss among this population. Barnet et al <sup>1</sup> using BAER identified 2.5% with severe (>60dB) bilateral loss at age 2 - 6 months. This suggests that the prevalence of clinically significant permanent hearing impairment among the Down's population could be in the order of 20 times greater than in the general population (1.1/1000 quoted by Bamford et al <sup>23</sup>). We therefore endorse the recommendation made by the working party for the National Deaf Children's Society (Quality Standards in Paediatric Audiology) that until universal neonatal screening is implemented babies with Down's syndrome should be included as a high risk group in targeted neonatal screening programmes <sup>14</sup>.

### Later infancy, childhood and adolescence

The major cause of the high prevalence (50 - 100%) of hearing impairment in this age group is conductive loss. This is usually secondary to serous otitis media (SOM - glue ear). Many children are already affected by Age 1 <sup>1,27</sup>. Schwartz and Schwartz <sup>18</sup> found otoscopic evidence of SOM in 59% of an unselected sample of children with Down's syndrome under age 11. The natural history does not proceed to resolution nearly as frequently as in the general population and for many chronic SOM with or without cholesteatoma formation persists into adolescence and adult life <sup>3,5,18,35</sup>. These same children may also have anatomical problems in the middle ear and sensorineural loss. The incidence of the latter increases with age and studies which span adolescent age groups suggest a prevalence around 20% as adult life begins <sup>4,25,28</sup>.

### *Preschool screening.*

It has been suggested that the only way to ensure appropriate diagnosis and treatment among this high risk group is for all children with Down's syndrome to be seen by secondary or tertiary level audiology and ENT services from an early age. There is evidence that this would indeed identify more of those with hearing impairment <sup>3</sup>. However there is no evidence as to whether this practice would confer greater long term benefit than initial screening at primary level, provided that such screening is available and subject to rigorous quality control. We do however have some evidence that when children with Down's syndrome go through primary health care screening for hearing loss so many are identified with possible problems and referred to specialist services that it could well prove more economic to see them all at secondary or tertiary level in the first place <sup>29</sup>. We urgently need more information about developmental outcomes for children with Down's syndrome who have gone through different screening/diagnostic protocols at different ages.

The children's subgroup of the National Screening Committee has now recommended that neonatal screening for the detection of permanent congenital hearing impairment (PCHI) should be universally adopted <sup>34</sup>. They recognise that as this is implemented the community based infant distraction test (IDT) is likely to be phased out and recommend that other surveillance measures should be set in place at local level. Neonatal screening for PCHI will detect only a

small proportion of those with Down's syndrome who are likely to develop hearing impairment in the first year because most hearing impairment in this group is due to early onset SOM (glue ear)

The committee also advise that if retained that the IDT screening threshold should be raised to 45 or 50dB. This threshold is likely to be too high to be useful for those with Down's syndrome. Furthermore detection of mild/moderate impairments for which these children do not easily compensate is not easy on behavioural grounds. Hence abolition of the first year community based IDT in its present form is likely to leave children with Down's syndrome at a disadvantage. In view of these changes and in the absence of firm evidence as to the efficacy of the present IDT as a screening tool for hearing impairment in this population we recommend that in future all in this high risk group are seen and monitored at secondary or tertiary level from the outset.

Bearing in mind variations in local provision as the new recommendations are implemented we have not made any recommendation as to how essential screening should be implemented but have specified those tests which are essential at each age. (see *diagnostic methods*) We have chosen 10 months as the target age by which every child with Down's syndrome should have been shown either to have unequivocally normal hearing or have been referred for full review at secondary or tertiary level. We believe that this target is reasonably and universally achievable within present surveillance capabilities.

#### *Screening for school age children*

We have already emphasised the high prevalence of SOM in this age group<sup>18</sup>. In addition it cannot be assumed that most of those apparently successfully treated in the preschool years need no further follow up. A recent audit in Leeds District<sup>30</sup> showed that 60% of children with Down's syndrome who had had previous surgical intervention for middle ear disease continued to have hearing loss at their most recent hearing test. This is one piece of evidence which underlies our insistence that surveillance should be ongoing for all children throughout childhood. Another of course is the progressive impact of sensorineural loss<sup>25</sup> as the children get older. We have no idea of the relative cost-effectiveness of surveillance at one, two or three yearly intervals. Until we have some hard data clinical experience suggests to us that two yearly is likely to be reasonable.

#### Adults

Adults with Down's syndrome have a high risk of progressive sensorineural loss. Some also have ongoing chronic SOM with or without cholesteatoma. The sensorineural loss appears to some extent to be a manifestation of precocious ageing. Presbycusis develops 30-40 years earlier than in the general population<sup>26</sup>. Shortened cochlear length may also be implicated. Evanhuis<sup>6</sup> found 57% of those age 35-62 years had bilateral loss greater than 40dB. In 68% the loss was sensorineural. Few practical or psychological problems were encountered in gaining the subjects' cooperation for this investigation. Prior to the study hearing loss had been diagnosed in only around 25% of those considered to have a handicapping loss. Evanhuis and others also emphasise that behaviours associated with undiagnosed progressive hearing impairment in adults with Down's syndrome may lead to a false diagnosis of dementia.

#### *Screening for adults*

The high incidence and incremental nature of hearing impairment among these adults, the possibility of mistaken diagnoses of dementia and the fact that these people are unlikely to volunteer information which suggests progressive or new loss leads to our recommendation of screening throughout life. Currently we do not have longitudinal information to guide us as to how frequently such screening should take place. We have therefore suggested that two yearly

testing continues into adult life. If this target is achieved it will provide information on which an evidence based recommendation for frequency of testing can be made.

### Diagnostic methods

In an appropriate setting, and with an experienced tester, it is reasonable to have relatively high expectations for many of these children as far as performance abilities are concerned (5 and Davies - personal communication). Most will respond to standard tests<sup>12</sup> appropriate to developmental age.

We have suggested that full audiological review should include impedance testing, audiological thresholds and otoscopic investigation. None of these procedures carried out in isolation can identify the whole range of possible pathologies<sup>18</sup>. Impedance testing does not identify sensorineural loss which is prevalent in this population. Both impedance testing and determination of auditory thresholds are affected by wax in the ear canals. In addition it is our consensus view that all people with Down's syndrome should be seen from time to time by an ENT specialist who is well informed about the syndrome. All, and particularly children, have a high incidence of problems related to the upper airways, middle ear and eustachian tubes all of which are secondary to underlying structural abnormalities in these areas. Many of these problems can be actively managed (eg persistent catarrh and upper airway obstruction) are interrelated and impinge directly on middle ear function. Hence a specialist overview is needed in addition to impedance and auditory threshold testing.

Compared with other areas there is a surprising amount of research evidence about the use of Auditory Brain Stem (ABR) responses in the investigation of hearing impairment in people with Down's syndrome<sup>6,22,36,37</sup>. All investigators caution about the interpretation of ABR findings in this population.

For the general population it is accepted that whilst hearing loss of certain types will reliably affect ABRs it is not safe to reason that an ABR abnormality indicates a hearing deficit<sup>37</sup>. For those with Down's syndrome the situation is further complicated because this group has unique ABR findings - in particular short central conduction times regardless of hearing status. This may make identification of hearing loss particularly difficult<sup>22,37</sup>. Widen<sup>22</sup> states 'If the ABR is to be used as a clinical tool to estimate hearing sensitivity of patients with Down syndrome, particular care should be taken to base interpretation on response detection level as well as latency because comparison of latency values to norms at suprathreshold levels may lead to false-negative findings'.

Social hearing loss may be overestimated by brainstem audiometry. A considerable part of speech information is expressed in frequencies lower than the ABR sensitivity threshold of 3kHz. Evanhuis<sup>6</sup> usefully compares Brain Stem Evoked Response Audiometry (BERA) and pure tone audiometry threshold findings in adults with Down's syndrome and finds both concordance and non- concordance.

The conclusion appears to be that if ABR is to be used as a clinical tool interpretation must be by someone who is aware of the unique abnormalities normally found in people with Down's syndrome. Data thus interpreted may indeed be useful in conjunction with other tests when trying to establish the hearing status of adults with Down's syndrome<sup>7</sup>. ABR should never be used as the sole investigation other than for newborns and for those who are so profoundly intellectually disabled as to be unable to cooperate usefully with other procedures. A study is currently underway as to whether incorporating ABR into a childhood screening/diagnostic programme gives useful additional information which may significantly improve outcome for children with the syndrome.

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