Original Article

High Prevalence of Hearing Loss in Down Syndrome at First Year of Life

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Abstract

Introduction: Infants with Down syndrome (DS) are at higher risk of hearing loss (HL). Normal hearing at one year of age plays an important part in language development. An audit was conducted to determine the impact of the newborn hearing screening program on the incidence, type and timing of diagnosis of HL during first year of life. Materials and Methods: Infants with DS were scheduled for Universal Newborn Hearing Screening (UNHS) within 4 weeks of life. If they passed, they had a high-risk screen at 3 to 6 months. They were referred to the otolaryngology department if they did not pass the UNHS or the high-risk screen. Information was obtained from the computerised data tracking system and case notes. Infants born from April 2002 to January 2005 and referred to the DS clinic of our hospital were analysed. Results: Thirty-seven (82.2%) of 45 infants underwent UNHS, of which 12 (32.4%) infants did not pass. Of remaining 33 infants, 27 had high-risk screen done of which 14 (51.8%) did not pass. Twenty-eight infants were referred to the ear, nose, throat (ENT) clinic: 12 from UNHS, 14 from high-risk screens and 2 from the DS clinic. Eleven (39.2%) defaulted follow-up. Fourteen (82.3%) of 17 infants who attended the ENT Clinic had HL. Twelve (85.7%) were conductive, and 2 (14.2%) mixed. Nine (64.2%) had mild-moderate HL and 3 (21%) had severe HL. The mean age of diagnosis was 6.6±3.3 months. All were treated medically, plus surgically if indicated. By 12 months of age, the hearing had normalised in 4 (28.6%) infants and remained the same in 3 (21.4%). Five (35.7%) defaulted follow-up. Thirty-five out of 45 (77.8%) underwent complete hearing screen in the first year of life (UNHS & High-risk screen). Six out of 45 (13.3%) had incomplete screening. Fourteen out of 41 (34.1%) had HL of varying degrees. Four out of 45 (8.8%) did not have any audiological assessment in first year of life. Conclusion: The incidence of HL in the first year of life was high (34.1%). Eighty-five percent were conductive with 64.2% in mild-moderate range. One third of infants hearing normalized after treatment, one third remained unaltered and one third of infants did not attend follow-up. An aggressive approach involving early screening after birth and continued surveillance and early referral to appropriate agencies are essential for establishing timely diagnosis and treatment. Measures to reduce the high default rate during long-term follow-up are needed. Parent education and integrated multidisciplinary follow-up clinic may be useful.

Ann Acad Med Singapore 2011;40:493-498

Key words: Deafness, Trisomy, UNHS

Introduction

Down syndrome (DS) is the most commonly occurring genetic abnormality involving approximately 1 in 700 births.[1] The increasing life expectancy of individuals with DS has revealed the presence of several unexpected pathological processes.[2] Otorhinolaryngological disorders hold an important place amongst them because of their high incidence and severity.[2] These frequently cause an increase in DS-related handicap. Studies have shown that even mild hearing loss (HL) of less than 15dBHL can adversely impact speech perception, learning, cognition and speech development.[3-8] This is particularly important in DS where expressive language skills lag behind cognitive abilities.[3,9] Early diagnosis and treatment of hearing loss has the potential to improve language abilities that may indirectly affect educational vocational costs and may increase lifetime productivity.[10]

It has been clearly shown that HL occurs more often in infants with DS than in healthy infants or those with other developmental disturbances.[11,12] It is caused by craniofacial, functional and immune system abnormalities.[2] Reported incidence in the literature ranges from 38% to 82%.[13-16]

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Therefore, it is of paramount importance that HL in infants with DS is diagnosed and treated early in life for normal speech with intelligible phonation.

Most authors recommend hearing screening of infants with DS early in the neonatal period to improve later learning and speech development.[3,17] The American Academy of Pediatrics (AAP)[18] has recommended that objective testing for hearing loss in DS should be done at birth or 3 months and that surveillance should continue during childhood. In their position statement on early hearing detection and intervention (EHDI), the Joint Committee on Infant hearing (JCIH)[19] recommends hearing screening by 1 month, comprehensive audiological evaluation by 3 months for those who do not pass the neonatal screen and intervention for confirmed hearing loss by 6 months of life. Aggressive, meticulous, complete diagnosis and treatment of ear diseases in DS starting soon after birth has shown a reduction in hearing loss to 2%.[3] Normal hearing at 1 year of age maximizes speech and language acquisition that has the potential to minimise personal-social maladjustments and emotional impairments in these children with DS.[20]

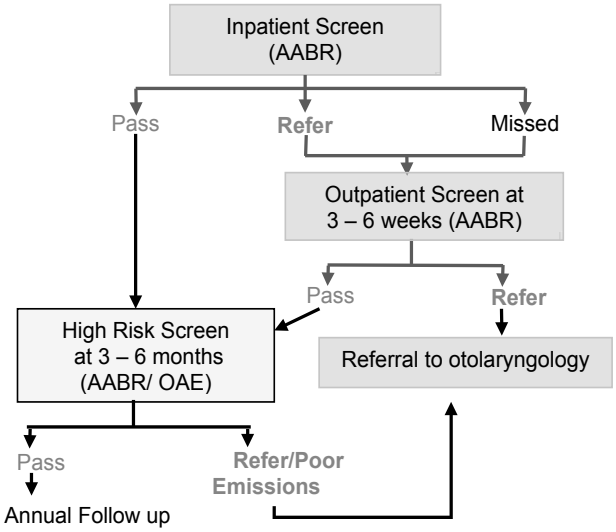

Fig. 1. Hearing screening protocol for infants with DS.

Objective

As part of an internal audit on the Universal Newborn Hearing Program and the ongoing hearing surveillance in high-risk populations, infants with DS were reviewed to

(i) determine the use of early hearing screening in infants,

(ii) determine the incidence, type, timing of diagnosis of HL during the first year of life as a result of routine hearing screening and the response to timely interventions.

Methods and Materials

In KK Women's and Children's Hospital (KKWCH), all newborns were screened for HL after birth using the Auditory Automated Brainstem Response (AABR). The results were expressed as "pass" or "refer". Infants at high-risk of HLs were rescreened at 3 to 6 months using the AABR or the Otoacoustic Emission (OAE). OAE results were expressed as "normal emissions" (or "pass") and "poor emissions". The screening protocol is shown in Figure 1.

In the AABR, infants needing >10,000 sweeps to obtain a pass result in any ear were considered to have abnormal results ("refer"). Patients with abnormal, "refer" or "poor emissions" in the UNHS or high-risk screen were referred to the Otolaryngology Department for further evaluation. They were objectively assessed using the OAE, Tympanometry and steady state evoked potential (SSEP). Hearing loss was defined as mild (26 to 40dbHL), moderate (41 to 60dbHL), severe (61 to 80dbHL) or profound (>80dbHL).[21] Interventions included removal of wax, antibiotics for otitis media or surgeries such as myringotomy, grommet insertion or both. Patients were re-evaluated subjectively by the treating physician and objectively with hearing screening whenever possible. Screening and subsequent hearing assessment data was prospectively entered into a computerized data management system called HiTRACK.

This paper is the result of a retrospective audit of a cohort of children with DS born between April 2002 and January 2005, who were followed up in the KKWCH's Down Syndrome Clinic. The study protocol was approved by Institutional Review Board. Hearing screening data was extracted from the HiTRACK system and details of audiological management were obtained from the case notes.

Results

Forty-five infants with DS were followed up in the clinic, including 5 outborn infants who were referred to DS clinic for further management. There were 31 males and 14 females. Figure 2 provides the overview of the patient flow in the study.

Figure 3 provides the outcome of hearing screening in these infants. Of the 8 infants who missed the UNHS, 5 were outborn and 3 were inborn. Parents of 3 inborn DS infants refused the UNHS. However, of these, 2 infants attended high-risk screening and 1 was lost to follow-up. Of 4 infants who did not have any hearing screening in the first year of life, one migrated to China (outborn), 2 were lost to follow-up (1 inborn, 1 outborn) and 1 (outborn) was screened after 12 months.

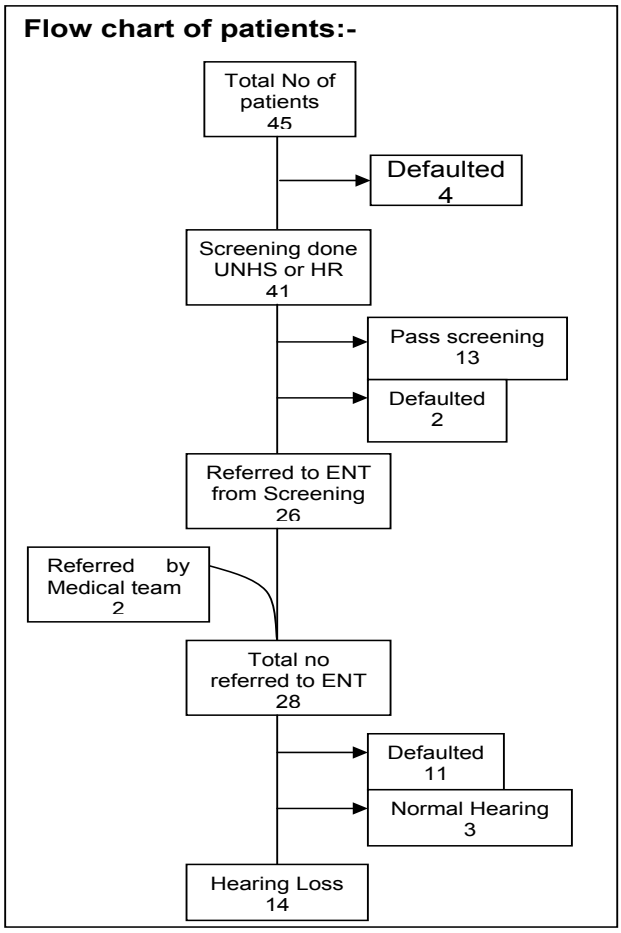

Fig. 2. Flow chart of patients.

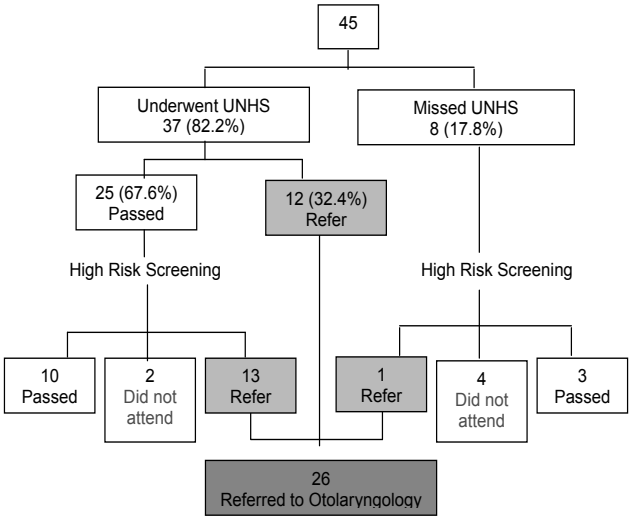

Fig. 3. Outcome of hearing screening in infants with Down Syndrome.

Table 1. Referral Source and Hearing Outcome

	UNHS	High-risk	Doctor
Referred to ENT	12	14	2
Diagnosed with HL	5	7	2
Type of HL			
Mild	1	2	1
Moderate	2	3	0
Severe	2	0	1
Mixed	0	2	0
Normal	1	2	0
Defaulted	6	5	0

Of 28 infants who were referred to the Department of Otolaryngology after the UNHS and high-risk hearing screening (26) and for medical indications (2), 17 (60.7%) were assessed for hearing loss. Fourteen out of 17 (82.3%) were diagnosed to have hearing loss. Three out of 14 (17.7%) had normal hearing. Eleven out of 28 (39.3%) did not complete their audiological assessment. Table 1 provides details about the outcome of referred patients.

Figure 4 provides details of outcome of HL in the 14 affected infants. The mean age of diagnosis was 6.6 ± 3.3 months. Of the 2 infants who had mixed HL, one was unchanged when reassessed at one year of age after a trial of antibiotics for otitis media. The other infant defaulted the follow up hearing assessment. Both of them had no other risk factor other. No further investigations were done for them during the first year of life.

Of 14 infants who had HL, 3 had history of admission to the Neonatal intensive care unit for respiratory distress after birth. All of them received Continuous positive pressure (CPAP) ventilation. These infants also received aminoglycosides for presumed sepsis and drug levels were normal. Two infants were re-admitted to hospital for acute respiratory infections during the first year and both of them were discharged after 48 hours of treatment. One infant attended the children's emergency for respiratory infection, he was treated and discharged well. Of the 5 infants who had moderate HL at one year of age, 2 remained unchanged, 1 had unilateral HL and was advised further follow-up. The other infant had bilateral moderate HL, was advised hearing aids but the parents were not keen for intervention.

Eleven infants did not attend their otolaryngology follow-ups by one year of age. Of these, 4 had medical and surgical problems with the cardiovascular and/or respiratory systems requiring multiple admissions to hospital. Two were on follow-up in the DS clinic but did not keep their otolaryngology appointments, including one child who lived outside of Singapore and returned infrequently for follow-up. Three were lost to follow-up, including one child who returned to China. The records of 2 infants were

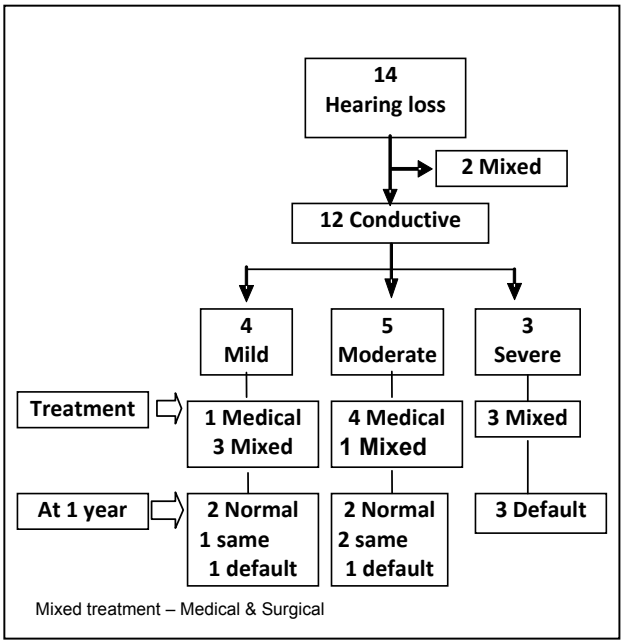

Fig. 4. Outcome of Infants with Hearing Loss.

not available.

Discussion

In this cohort of infants with DS, 91.1% underwent objective hearing screening in the first year of life, with 82.2% being screened during their birth admissions. Prior to the launch of the UNHS in KKWCH in 2002, only 10.6% of infants with DS were screened in the first 3 months, with 34.7% screened by the end of the first year of life (Down syndrome clinic, Department of Neonatology 2005, unpublished data). The literature testifies that for many years, hearing evaluation in DS was very difficult and unreliable.[14] Precise objective hearing assessment regardless of age or mental condition became possible only after the introduction of testing brain auditory evoked potentials in clinical practice.[22] Thus, by screening all infants through the UNHS program, infants with DS who are at high risk for hearing loss were screened far more comprehensively than ever before.

Our attempts to adhere to the recommendations of the AAP[18] and JCIH[19] in KKWCH allowed screening during the birth admission in 37 (92.5%) of 40 inborn infants, which was close to the JCIH benchmark of screening 95% by 1 month of age. Of 44,465 infants, 99.8% were screened during the birth admissions during 2002 to 2005.[23] Those who missed the newborn screening either refused the hearing screening or migrated to other countries.

The AAP guidelines on health supervision of DS and many other authors[18,24,25] recommend continued surveillance for HL at each follow-up visit in this high-risk group. In KKWCH, as part of continued surveillance, a second hearing screen (referred to as a "high-risk" screen) is scheduled between 3 to 6 months of life in those who passed the first newborn hearing screen. Eighty-two percent (27 of 33 eligible infants) underwent objective high-risk hearing screening. Fourteen infants were referred for formal audiological assessment, facilitating the early diagnosis of HL in 6. Overall, 69.7% (23/33) underwent both UNHS and high-risk screening.

HL was diagnosed before the end of the first year of life in 34.1% of who had at least one hearing screen and in 82% (14/17) of those who underwent audiological evaluation. There are few papers that have reported early hearing outcomes in DS for comparison. However, Hess et al[26] reported similar results in older children of 4.6 ± 3.4 years with DS. Twelve of 14 (85.7%) of patients had treatable and potentially correctable conductive HL that was mild to moderate in severity in 9 (64.2%). This is important as a mild to moderate HL loss can also negatively affect the speech and language development even in normal children.[2-5,15,16] Recently, McPherson et al[27] reported similar findings in the severity of conductive hearing loss in school children with DS at 10 to 12 years. Sensorineural hearing loss was uncommon in our cohort of DS infants, which Hess found in 8.6% of his cohort. This difference in pattern is surprising and raises the question that sensorineural hearing loss may be a late occurring problem in DS children. This can be result of the early onset of presbycusis and cochlear outer hair cell dysfunction.[28] As it is not very clear from the present literature when sensorineural HL becomes predominant, further studies are required to establish this. The current literature does show that the HL in DS that starts in early infancy and continues in early school age is mainly conductive, which has good potential for treatment. An aggressive approach creates the opportunity for improvement of language acquisition in these infants. A better understanding of the onset and severity of sensorineural HL in school age will reveal the window of opportunity, if any, after resolution of conductive HL to maximise the language learning in early childhood.

The mean age of diagnosis of HL in our study was 6.6 ± 3.3 months. This was a significant improvement over the pre-UNHS era (before 2002) where the mean age of diagnosis in KKWCH was >24 months. (Unpublished data from the department of Neonatology, KKWCH). Hess et al[26] and McPherson et al[27] reported diagnosis of HL at 4.6±3.4 and 10.2 ± 2.4 years respectively (Table 2). The early diagnosis of hearing loss allows early intervention. Six of our patients received only medical treatment for middle ear infection or wax, but the majority also needed surgical interventions such as myringotomy and grommet insertion. In McPherson's[27] study where hearing loss was diagnosed at 10 to 12 years, intervention was confined to

classroom modification and sound amplification. It would be interesting to see if early medical and surgical intervention in the first year of life reduces the eventual need for such later interventions.

At one year of age, one third of our affected infants had improved, one third of conductive HL remained unchanged and one third was not assessed. The incidence of HL at end of the first year of life after intervention in our cohort had fallen from 82.3% to 34.1%, which is keeping with the present literature.[3-5,9,28-31] If these patients had been undiagnosed and untreated, hearing-related handicap in these vulnerable patients with DS would have increased. The improvement in hearing status after treatment in one third of these infants is an encouraging finding, as Harigai et al[20] has shown that appropriate medical and surgical care causing improved hearing acuity, had a positive impact on human relationship and accelerated speech development. Shott et al[3] has shown that after eliminating causes of easily reversible hearing loss and with aggressive treatment of chronic ear disease, the residual HL was only 2%.[3] This suggests that there is a place for far more aggressive treatment of hearing loss than what was practiced in KKWCH.

In spite of having a formal hearing-screening program, the advantage of hearing assessment was limited to those who remained in follow-up. Other authors have also noted very high default rates in this high-risk group. Seventy-five percent defaulted diagnostic evaluation in McPherson's study[27] Possible cause of non-compliance with screening may have been the presence of other medical issues e.g. repeated chest infections (as in 2 of the patients in this cohort who were admitted twice during the first year or the need for multiple follow-up appointments for other medical or surgical problems associated in DS). Several of our patients had cardiac defects and surgical issues, including duodenal atresia, Hirschprung's disease, respiratory infection (2 infants were readmitted for respiratory infections). It is then likely that in such cases, hearing assessment follow-up is a low priority for parents. Several of our infants could not be formally assessed with sedation. Parents were often reluctant to allow their infants to be anaesthetised for hearing assessment. Parental awareness and perception of hearing loss as noted by McPherson et al[27] plays an important role in the audiological rehabilitation of these infants with DS.

Table 2. Comparison between our results and those from studies by Hess et al[26] and McPherson et al[27]

Variables studied	KKWCH	Hess et al[26]	McPherson et al[27]
Age of diagnosis	6.6 ± 3.3 months	4.6 ± 3.4 years	10 to 12 years
Incidence of HL	82%	50%	73%
Type of HL (conductive)	86%	82%	25%
Severity (Mild-Moderate)	57%	Not mentioned	60%
Treatment	Medical + surgical	Not mentioned	Non medical (Classroom modification, sound amplification)

Limitations of this study

In this cohort, there was a 40% default rate in patients who were referred to the Otolaryngology department. Thus, the results may not be representative of the whole group of patients with DS. The hearing assessment in 2 patients at the end of the first year was evaluated clinically by treating physicians with no formal testing done.

Conclusion

The incidence of HL in DS is very high during the first year of life. In the majority of cases, it is mild to moderate in range and is conductive in nature. In one third of infants, HL normalised with treatment, one third remained the same and one third did not attend follow-up. Although the use of routine hearing screening after birth was far greater than the pre-UNHS periods, there was a significant default rate.

Recommendation

An aggressive approach involving hearing screening after birth, continued surveillance, appropriate referral for further investigation, diagnosis and management of HL is an important part of the high risk care of this special group of infants. Measures to reduce the high default rate need attention. Parent education regarding the importance of proactive measures to improve the hearing and integrated multidisciplinary follow-up clinic may be useful.

Why this study?

1. Previous studies have reported a high incidence of HL in children with DS and a mean age of detection from 4.6 to 12 years.[28]
2. There is a paucity of data on early detection and interventions of HL in infants with DS since the introduction of the EHDI program.
3. Normal hearing is very important in order to optimise

the potential of children with DS for acquiring early language skills.

What this study adds?

1. After the introduction of the UNHS program, 82.2% of DS infants underwent hearing screen and HL was diagnosed at a mean of 6.6 ± 3.3months.

2. The majority (86%) had conductive hearing loss (with the potential for intervention) of mild-moderate severity in more than half (57%).

3. One third of those who remained on follow-up had normal hearing after treatment at one year of age.

4. One third did not complete their audiological assessment. Despite an established hearing screening program and the long-term implications of hearing loss, hearing assessment remains a low priority for many parents of DS infants during the first year of life.

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