The Prevalence of Congenital Hearing Loss in Neonates with Down Syndrome

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Objective To determine the prevalence of hearing loss in newborns with Down syndrome.

Study design We performed a cross-sectional, retrospective chart review of all infants with Down syndrome born at a university-affiliated hospital (n = 77) or transferred in to the associated pediatric hospital (n = 32) following birth at an outlying hospital between 1995 and 2010. We determined the rate of failure of newborn hearing screens, the proportion of infants lost to follow-up, and the rate of confirmed hearing loss, as well as the associations of risk factors for hearing loss with confirmed hearing loss.

Results Of the 109 patients with hearing screening data, 28 failed their newborn hearing screen. Twenty-seven infants were referred for audiologic evaluation, and 19 completed the evaluation. Fifteen of these 19 infants (79%) had confirmed hearing loss. The prevalence of congenital hearing loss in this sample of neonates with Down syndrome was 15%. Exposure to mechanical ventilation was the sole known risk factor associated with hearing loss. In this study, the loss to follow-up rate for infants with positive hearing screens was 32%.

Conclusion Newborns with Down syndrome have a higher prevalence of congenital hearing loss compared with the total neonatal population (15% vs 0.25%). Continued monitoring of hearing is needed in children with Down syndrome. *(J Pediatr 2015;166:168-71)*.

Down syndrome is one of the most common chromosomal abnormalities in the US, affecting 1 in 691 births and resulting in approximately 6037 infants born with Down syndrome each year.[1] Previous studies have reported a prevalence of hearing loss among children and adolescents with Down syndrome ranging between 45% and 90%,[2-10] compared with 2.5% in the general pediatric population.[11] The variability in prevalence data is likely secondary to the use of small convenience samples, variable hearing testing modalities, and a lack of standardized criterion defining "abnormal" hearing.

Individuals with Down syndrome are prone to otolaryngologic anomalies that complicate the diagnosis and classification of hearing impairment. Evaluating the tympanic membrane in young children with Down syndrome is challenging secondary to stenosis of the external auditory canal and increased rates of cerumen impaction.[9] Middle ear abnormalities, including eustachian tube dysfunction, remnant tissue in the middle ear cavity, and ossicular malformations, are frequently seen and further contribute to the higher rate of conductive hearing loss reported in this population.[4,8,9] Inner ear hypoplasia and dysplasia of the cochlea, cochlear nerve canal, lateral semicircular canal, and cochleovestibular nerve are hypothesized to explain the frequent and progressive nature of sensorineural hearing loss seen in individuals with Down syndrome.[12] Studies of adolescents and adults with Down syndrome have demonstrated an increasing prevalence of sensorineural hearing loss with age.[3]

Although numerous studies have confirmed the increased incidence of hearing loss in individuals with Down syndrome, the prevalence of congenital hearing loss had not been studied previously. Hearing loss in infants has been associated with lifelong deficits in speech and language acquisition, poor academic performance, personal-social maladjustments, and emotional difficulties.[13-15] Early identification of infants with hearing loss may help mitigate the adverse consequences associated with hearing loss.[16] The institution of universal newborn hearing screening provides a new opportunity to examine the prevalence of congenital vs acquired hearing loss in the population of children with Down syndrome.

The aim of the present study was to collect and analyze the data obtained from universal newborn hearing screening and subsequent audiologic evaluations to establish the prevalence of congenital hearing loss in neonates with Down syndrome. We also examined the referral rate for follow-up of abnormal hearing screening, the proportion of infants lost to follow-up, and the associations of risk factors for hearing loss with confirmed hearing loss.

AABR	Automated auditory brainstem response
NICU	Neonatal intensive care unit

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Methods

Following Institutional Review Board approval, a retrospective chart review was performed of all infants with Down syndrome born at University Hospitals in Cleveland or transferred in following birth at an outlying hospital between March 8, 1995, and December 17, 2010. Deidentified data, including newborn hearing screening results, risk factors associated with hearing loss, referral to audiology, and results of subsequent audiologic evaluations, were collected.

The hearing loss risk factors assessed included neonatal intensive care unit (NICU) admission for ≥5 days, maternal prenatal illness (ie, documented infections associated with hearing loss to include cytomegalovirus, herpes, syphilis, rubella, and toxoplasmosis), family history of hereditary childhood hearing loss, low Apgar scores (0-4 at 1 minute or 0-6 at 5 minutes), birth weight <1500 g, presence of a syndrome associated with hearing impairment, indirect bilirubin >20 mg/dL, exchange transfusion, meningitis, craniofacial anomalies, exposure to ototoxic drugs, and mechanical ventilation. These risk factors were initially selected from the 1994 Position Statement of the Joint Committee on Infant Hearing and modified after subsequent revisions.

The screening protocol and tools did not change during the 15-year study period. All infants born at or admitted to University Hospitals in the neonatal period underwent universal newborn hearing screening using automated auditory brainstem response (AABR) technology. If an infant received a nonpass result (referred) during the initial screen, then a second AABR was attempted before discharge. Infants who did not pass the hearing screening after 2 AABR screens were referred to audiology for further evaluation, which included auditory brainstem response testing, distortion product otoacoustic emissions testing, tympanography, and bone and air conduction studies. Infants identified as at increased risk for hearing loss (based on the risk factors listed above) underwent an audiologic evaluation before discharge. Lower-risk infants were scheduled for outpatient audiologic evaluation on discharge to home.

Association of known risk factors for hearing loss with the presence of confirmed hearing loss were analyzed using the Fisher exact test. $P \leq .05$ was considered to indicate statistical significance.

Results

Of the 131 infants who met the inclusion criteria, 126 had charts available for review. The remaining 5 infants' charts could not be located by the medical records department. Six infants died before screening, and 11 infants did not have a documented hearing screen, resulting in a study population of 109 infants (57 males). Of the 109 infants included in this study, 77 were born at University Hospitals and 32 were transferred in from outlying community hospitals after birth.

Twenty-eight infants (25.7%) failed the newborn hearing screen (**Figure**; available at www.jpeds.com). Twenty-seven infants were referred for follow-up audiologic evaluation, and 19 infants completed the evaluation. Fifteen of the 19 infants with audiologic data (78.9%) had confirmed congenital hearing loss, including 4 with unilateral hearing loss and 11 with bilateral hearing loss. The screening process only determined a pass/fail result, and the diagnostic evaluations did not consistently distinguish between conductive and sensorineural hearing loss.

In this population of newborns with Down syndrome, the prevalence of congenital hearing loss was determined to be 15%, based on 15 infants with confirmed hearing loss in a population of 100 (after having subtracted the 9 infants lost to follow-up). Given the 32% loss to follow-up rate and our concern that excluding these infants could have underestimated or overestimated the true prevalence of hearing loss, we calculated an estimated prevalence. Based on the high degree of agreement between failed AABR screening and confirmed hearing loss demonstrated on audiologic testing (78.9%), we estimated that an additional 7 infants with hearing loss would be identified in the lost to follow-up group (0.79 × 9 infants lost to follow-up). We determined an estimated prevalence of hearing loss in newborns with Down syndrome of 20% after adding the estimated 7 additional cases to the known 15 cases, resulting in 22 total cases of hearing loss in a population of 109 infants. If all of the children lost to follow-up had normal hearing, then the prevalence of congenital hearing loss would be 13.8%; if all had hearing loss, then the prevalence of hearing loss would be 22%.

Associations between known risk factors for hearing loss and presence of hearing loss were examined using the Fisher exact test, owing to the low frequency of some risk factors in the study population (**Table**). There were only 2 infants with documented maternal illness during pregnancy and 1 infant each with family history of hearing loss, bilirubin level >20 mg/dL, and meningitis. No infants received exchange transfusions for an elevated bilirubin level, and thus we did not include this risk factor in our calculations.

We compared the presence of risk factors for hearing loss in infants with confirmed hearing loss and infants who passed newborn hearing screening or had normal audiologic evaluations. The sole risk factor identified as positively correlated with the presence of hearing loss was mechanical ventilation

Table. Associations between risk factors for hearing loss and presence of hearing loss

Risk factors	Fisher exact test	Infants with risk factor, n (%)
NICU stay >5 days	0.159	77 (70.6)
Maternal illness	1.000	2 (1.8)
Family history of hearing loss	1.000	1 (0.9)
Low Apgar score	0.279	17 (15.6)
Low birth weight	0.097	8 (7.3)
Bilirubin >20 mg/dL	1.000	1 (0.9)
Meningitis	0.150	1 (0.9)
Defects of face/head	1.000	5 (4.6)
Ototoxic drugs (prenatal and postnatal)	0.101	53 (48)
Mechanical ventilation	0.008	19 (17.4)