

# Hearing Screening in Children With Skeletal Dysplasia

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**Objective:** To determine the prevalence of hearing loss and abnormal tympanometry in children with skeletal dysplasia.

**Design:** Clinical screening program.

**Setting:** National convention of the Little People of America.

**Patients:** Convenience sample of volunteers aged 18 years or younger with skeletal dysplasias.

**Interventions:** Hearing screening with behavioral testing and/or otoacoustic emissions, otoscopy, and tympanometry.

**Main Outcome Measures:** A failed hearing screen was defined as hearing 35 dB HL (hearing level) or greater at 1 or more tested frequencies or by a “fail” otoacoustic emissions response. Types B and C tympanograms were considered abnormal.

**Results:** A total of 58 children (aged  $\leq 18$  years) with skeletal dysplasia enrolled, and 56 completed hearing

screening. Forty-one children had normal hearing (71%); 9 failed in 1 ear (16%); and 6 failed in both ears (10%). Forty-four children had achondroplasia, and 31 had normal hearing in both ears (71%); 8 failed hearing screening in 1 ear (18%), and 3 in both ears (7%). Tympanometry was performed in 45 children, with normal tympanograms found in 21 (47%), bilateral abnormal tympanograms in 15 (33%), and unilateral abnormal tympanograms in 9 (20%). Fourteen children with achondroplasia had normal tympanograms (42%); 11 had bilateral abnormal tympanograms (33%); and 8 had unilateral abnormal tympanograms (24%). For those children without functioning tympanostomy tubes, there was a 9.5 times greater odds of hearing loss if there was abnormal tympanometry ( $P=.03$ ).

**Conclusions:** Hearing loss and middle-ear disease are both highly prevalent in children with skeletal dysplasias. Abnormal tympanometry is highly associated with the presence of hearing loss, as expected in children with eustachian tube dysfunction. Hearing screening with medical intervention is recommended for these children.

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**C**HILDREN WITH SKELETAL dysplasias have a number of otolaryngologic issues, including obstructive sleep apnea, nasal obstruction, airway management issues during general anesthesia, middle-ear disease, and hearing loss. While hearing loss is common in these children, the literature that demonstrates this consists largely of studies of patients with skeletal dysplasia who have presented for medical care. To our knowledge, there are no studies that have investigated hearing loss in skeletal dysplasia using testing outside the clinical domain.

While hearing screening of neonates is nearly universal in the United States, follow-up hearing assessment later in childhood is not. Hearing screening programs for children can identify children at risk for hearing loss, and such programs have been documented to improve audiologic follow-up and treatment of hearing loss in school-aged chil-

dren.<sup>1</sup> In addition, the Joint Committee on Infant Hearing<sup>2</sup> 2007 statement notes that children with craniofacial abnormalities are at risk for delayed-onset hearing loss, supporting aggressive sequential hearing assessments in the skeletal dysplasia populations. Children with achondroplasia appear to have verbal comprehension deficits that may be related to middle-ear disease and conductive hearing loss.<sup>3</sup>

We performed a hearing screening program at a national meeting of individuals with short-stature skeletal dysplasias. We sought to identify children with hearing loss or at risk for hearing loss to recommend appropriate follow-up evaluation and treatment. We attempted to measure the prevalence of hearing loss and middle-ear dysfunction in this population. With a hearing screening program at a convention for individuals with short-stature skeletal dysplasia, we studied the feasibility of such evaluation in a nonclinical setting. These

**Table 1. Demographics of Children With Skeletal Dysplasias**

Diagnosis	No.	Age, Mean (SD), y	Boys	Girls
Achondroplasia	44	7.8 (4.8)	26 <sup>a</sup>	17 <sup>a</sup>
Hypochondroplasia	1	6	0	1
Spondyloepiphyseal dysplasia congenita	2	7.5 (3.5)	0	2
Pseudoachondroplasia	1	2	1	0
Albright hereditary osteodystrophy	1	7	0	1
Dyggve-Melchior-Clausen	1	11	0	1
Cartilage hair hypoplasia	2	14.0 (1.4)	1 or 2 <sup>a</sup>	0 or 1 <sup>a</sup>
Primordial Dwarfism	3	7.3 (2.1)	2	1
Morquio	2	10.0 (1.4)	1	1
Spondylometaphyseal, Jansen	1	4	1	0
<b>Total</b>	<b>58</b>	<b>7.9 (4.6)</b>	<b>32<sup>a</sup></b>	<b>24<sup>a</sup></b>

<sup>a</sup>Sex was not specified for 1 child with achondroplasia and 1 child with cartilage hair hypoplasia.

screening programs are valuable for at-risk individuals who do not have access to medical care or who are unaware of the need for screening.

## METHODS

A convenience sample of individuals with skeletal dysplasias were recruited for hearing assessment with posted advertisements at the national meeting of the Little People of America in Nashville, Tennessee over 4 consecutive days in July 2010. Meeting attendees of all ages were eligible for hearing screening. Hearing screening was performed in 2 hotel rooms. The screening protocol was approved by the institutional review boards of the Alta Bates Summit Medical Center in Berkeley, California, and the Johns Hopkins Medical Institutions in Baltimore, Maryland. Written informed consent was obtained from parents and guardians prior to initiation of the screenings.

Hearing assessments were performed by audiology and speech and language pathology graduate students under the direction of 2 state-licensed and ASHA-certified (American Speech-Language-Hearing Association) audiologists. Individuals underwent hearing screening using MAICO MA 40 and MA 41 screening audiometers (MAICO Diagnostics), and tympanometry was performed with the Madsen Otoflex 100 (GN Otometrics) or the Grason-Stadler GSI 39 Auto Tymp tympanometers (Grason-Stadler). Otoscopy was performed by 1 of 2 board-certified otolaryngologists using Welch Allyn (Welch Allyn) handheld otoscopes. Otoacoustic emission testing was performed in 12 children using the Grason-Stadler GSI 70 Automated OAE device, principally for those too young to complete behavioral screening.

Hearing screening was performed using standard methods using air conduction only at 500 Hz, 1000 Hz, 2000 Hz, and 4000 Hz. A *pass* was defined as 2 appropriate responses to each presented stimulus below 35 dB HL (hearing level). Early in the screening process it became clear that the 500-Hz responses were not reliable, with multiple fails, perhaps because of ambient noise in the testing environment. Therefore, a *failed hearing screen* was defined as failure to respond at 1 or more of the frequencies 1000 Hz, 2000 Hz, or 4000 Hz in 1 or both ears below 35 dB HL, or by a “fail” response on automated otoacoustic emission testing.

Tympanometry results were recorded as type A, type A<sub>s</sub>, type A<sub>D</sub>, type B, or type C. For our analysis, all type A tympanograms were considered normal regardless of peak height. Type B and C tympanograms were considered abnormal, indicative of eustachian tube dysfunction. Tympanogram results were recorded for all patients who underwent such testing, even if tubes were present on examination. For 1 analysis of the relationship of abnor-

mal tympanometry and failed hearing screen, patients with indwelling tympanostomy tubes were excluded from analysis.

Patients also had otoscopy and completed a 12-item questionnaire that documented patient age and skeletal dysplasia diagnosis, briefly described hearing and otologic health, and reported any history of ear surgery and use of hearing aids. Children who failed hearing screening and their parents were advised to seek comprehensive medical and audiologic assessment of hearing to confirm and quantify the suspected hearing loss and to provide appropriate diagnosis and treatment.

Hearing status and tympanometry results (normal in both ears, abnormal in 1 ear, or abnormal in both ears) were examined in the screened population. Data analysis was performed using STATA, version 10 (StataCorp LP). The risk of hearing loss in relation to tympanometry results, in the entire cohort and after excluding those with functioning tympanostomy tubes, were calculated using  $\chi^2$  tests ( $P < .05$ ).

## RESULTS

Fifty-eight of 110 subjects who enrolled in this study were children (mean [SD] age, 7.9 [4.6] years; age range, 10 months to 18 years), and 56 completed the hearing screening. Two children with achondroplasia did not complete the hearing assessment owing to non-compliance related to age. Of those who completed the screening, 42 children had achondroplasia, and 14 children had 1 of 9 other skeletal dysplasia diagnoses (**Table 1**). Nine children had tympanostomy tubes in 1 or both ears on examination. Twelve children had otoacoustic emission testing as part or all of their testing algorithm.

Forty-one children had normal hearing as defined by our pass criteria (71%); 9 children failed in 1 ear (16%); and 6 children failed in both ears (10%). Thus, 15 children failed hearing screening in 1 or both ears (26%). Thirty-one of 44 children with achondroplasia passed the hearing screening (71%): 8 of these children failed in 1 ear (18%), and 3 failed in both ears (7%) for a total of 11 children with achondroplasia failing in 1 or both ears (25%). Over one-half of the children with tympanostomy tubes present in at least 1 ear on examination failed hearing screening in 1 or both ears, while only 20% of those without tubes failed in 1 or both ears ( $n=9$ ). **Table 2** summarizes the hearing screening data.

Tympanometry was performed successfully in 45 children: 21 had normal tympanograms in both ears (47%);

**Table 2. Hearing Screening Results for Children With Skeletal Dysplasias<sup>a</sup>**

Clinical Condition	Patients, No.	Pass Both Ears	Fail 1 Ear	Fail Both Ears	Fail 1 or Both Ears	Not Tested
Total skeletal dysplasia	58	41 (70)	9 (16)	6 (10)	15 (26)	2 (3)
Achondroplasia	44	31 (71)	8 (18)	3 (7)	11 (25)	2 (45)
Tympanostomy tube(s) present in 1 or both ears	9	3 (33)	3 (33)	2 (22)	5 (56)	1 (11)
No tubes present	49	38 (78)	6 (12)	4 (8)	10 (20)	1 (2)

<sup>a</sup>Unless otherwise indicated, data are given as number (percentage) of patients.

**Table 3. Hearing Screening and Tympanometry**

Tympanogram Result	Ears, No.	Passed Hearing Screen	Failed Hearing Screen	No Hearing Assessment
A	51	46 (90)	4 (8)	1 (2)
B	30	20 (67)	7 (23)	3 (10)
C	9	7 (78)	2 (22)	0 (0)

<sup>a</sup>Unless otherwise indicated, data are given as number (percentage) of ears.

15 had abnormal tympanograms in both ears (33%); and 9 had an abnormal tympanogram in 1 ear only (20%). Fourteen children with achondroplasia had normal tympanograms (42%), while 11 had abnormal tympanograms in both ears (33%), and 8 had abnormal tympanograms in 1 ear only (24%). **Table 3** lists the hearing screening results for each tested ear by tympanogram result. While fewer than 8% of ears with type A tympanograms failed screening (4 of 51), ears with type B or type C tympanograms had similar failed screening rates, 23% (7 of 30) and 22% (2 of 9), respectively.

Odds ratios were calculated to examine the relationship between abnormal tympanometry and hearing screening results in this cross-sectional study. There was a 5.4 times greater odds of hearing loss if there was abnormal tympanometry ( $P = .04$ ). We reasoned that some children with patent tympanostomy tubes would be considered to have abnormal (flat) tympanograms based on our criteria, even though they had treated middle-ear disease. We did additional analysis by excluding children with patent tympanostomy tubes on examination. For those without tubes, there was a 9.5 greater odds of hearing loss if there was abnormal tympanometry ( $P = .03$ ). In the total population of children with achondroplasia both with and without tubes, there was a 7.1 greater odds of abnormal hearing if tympanometry was also abnormal, but this did not reach statistical significance ( $P = .06$ ).

While over one-quarter of children failed hearing screening in 1 or both ears, responses on the questionnaire indicated that only 1 child used hearing aids, and this same child was the only one previously told that hearing aids were needed.

#### COMMENT

Ear disease and hearing loss are common in patients with skeletal dysplasia. Glass et al<sup>4</sup> reported audiologic testing in 38 patients with skeletal dysplasia, and 27 of these patients were 20 years or younger. Seventeen of 28 patients with achondroplasia had hearing loss in at least 1

ear (61%), using a 20-dB threshold for normal hearing. About two-thirds of the patients with hearing deficit had conductive hearing loss. Only 2 of their 10 patients with other types of skeletal dysplasias had hearing loss, 1 with conductive and 1 with sensorineural hearing loss. Stura et al<sup>5</sup> studied the hearing of patients with skeletal dysplasias, all 21 years or younger, and found hearing loss in 10 of 18 patients with achondroplasia (56%), 7 with conductive hearing loss and 3 with sensorineural loss. Two of 10 patients with other types of skeletal dysplasia had hearing loss (20%). The threshold used to define hearing loss was not reported in the article, however.

Berkowitz et al<sup>6</sup> retrospectively reviewed the otologic histories of 61 patients with achondroplasia who were younger than 20 years. Fifty-four percent had tympanostomy tubes inserted at least once at a mean age of 38 months. Collins and Choi<sup>7</sup> retrospectively reviewed the charts of 22 patients with achondroplasia. Fifteen had documented otologic disease, and 7 patients had a total of 19 audiologic tests available for review. Thirteen of 19 audiograms showed conductive hearing loss in at least 1 ear (68%), and 1 patient had sensorineural hearing loss. No record of hearing aid use was found for any patient.

Our hearing screening program identified potential hearing loss in over a quarter of children who completed testing, and middle ear disease in over half of these children. Abnormal tympanometry and/or the presence of tympanostomy tubes in 1 or both ears was associated with failed hearing screen, as would be expected for a population where middle-ear dysfunction was the likely cause of hearing loss. Achondroplasia is the most common skeletal dysplasia, and indeed 44 of 58 children enrolled in this hearing screening had this diagnosis (76%). While hearing loss and middle-ear disease is quite common in achondroplasia in this study and in the literature, we cannot quantify the association of hearing loss and/or middle ear disease with other skeletal dysplasia diagnoses because the number of patients with such diagnoses is small. In our series, 2 of 2 children with spondyloepiphyseal dysplasia congenita failed hearing screen-

ing in both ears; 1 of 2 children with Morquio syndrome failed in both ears; and 1 of 2 children with cartilage hair hypoplasia failed in 1 ear.

To our knowledge, the present report describes the largest study of hearing health in children with skeletal dysplasia. We confirmed that hearing loss was highly prevalent in this group of patients and that hearing screening is feasible even in a nonclinical setting such as this national meeting of individuals with short-stature skeletal dysplasias. Such hearing screening is valuable for the skeletal dysplasia population because the presence of hearing loss seems to be underappreciated and interventions for hearing loss underutilized, at least by patient self-report. Our data support the recommendation that children with achondroplasia, and perhaps with selected other skeletal dysplasias, have hearing screening or formal testing yearly starting at age 12 months if not sooner.<sup>8</sup>

The present study, as a pilot and screening study, has several limitations. First, this cross-sectional study determined the prevalence of hearing loss simultaneously with that of abnormal tympanometry. Without a longitudinal study of a defined skeletal dysplasia cohort, we cannot conclude a temporal or causal relationship between abnormal tympanometry and hearing loss. In addition, selection bias in the tested population is possible with volunteer recruitment through advertisement at a busy national meeting. Patients with known hearing loss may defer such screening, as they presumably have already been diagnosed and hopefully treated. Conversely, patients who do not suspect any hearing loss may defer such screening. Our selection of a better-than 35-dB threshold for "pass" is less stringent than many hearing screening protocols. Newborn hearing screens usually use a 35- to 40-dB pass criterion, but such programs are aimed at identifying moderate to severe congenital sensorineural hearing loss.<sup>9</sup> Most hearing screens of school-aged children use a 20- to 25-dB pass criterion because they are trying to identify even mild hearing losses that may be conductive or sensorineural in origin.<sup>10</sup> The testing environment for this program was not acoustically ideal, leading to the decision to use a 35-dB threshold to identify individuals with moderate or severe hearing loss. It is likely that we have underestimated the incidence of hearing loss in children with skeletal dysplasias, even though more than a quarter of the children tested had what appeared to be at least moderate hearing loss in 1 or both ears.

Future studies of hearing and otologic health in children with skeletal dysplasias should include uniform evaluation of a large group of affected individuals to include diagnostic otoscopy, full air- and bone-conduction audiometry, impedance audiometry in a sound-controlled environment, and, where indicated, binocular microscopic evaluation of the ears and/or temporal bone imaging. Additional hearing screening programs in a nonclinical setting such as the Little People of America meeting should include recruitment efforts to avoid selection bias; optimization of the testing environment; and testing at 20- 25-dB threshold levels if possible to identify all children with hearing loss.

In conclusion, hearing loss and middle-ear disease are common in children with skeletal dysplasias. Abnormal

tympanometry findings appear to predict a failed hearing screen, as would be expected in this population of children where eustachian tube dysfunction and resultant conductive hearing loss are the most significant factors affecting hearing. Hearing screening is feasible at national meetings of patient groups in a nonclinical setting.

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