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Hearing Loss in Oto-spondylo-megaepiphyseal Dysplasia (OSMED): Case Studies

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Abstract

Oto-spondylo-megaepiphyseal dysplasia (OSMED) is considered to be an autosomal recessive disorder characterized by sensorineural hearing loss, short extremities in spite of normal body length, and abnormally thick joints. We present audiologic results and follow-up of two siblings (normal mother, affected father, 3 of 4 affected children). Given the severity of the hearing loss and, in some cases, severe myopia, early audiologic intervention is highly desirable.

Key Words: Hearing loss, oto-spondylo-megaepiphyseal dysplasia (OSMED), skeletal dysplasia

Oto-spondylo-megaepiphyseal dysplasia (OSMED) is considered to be an autosomal recessive disorder characterized by short extremities in spite of normal body length, abnormally thick joints, and sensorineural hearing loss (Giedion et al, 1982). Although Giedion et al (1982) reported a narrow internal auditory canal in one case, the mechanism of hearing loss in OSMED has not been clearly identified. Other reported manifestations including cleft palate or bifid uvula and severe myopia have led investigators to believe that OSMED and skeletal dysplasias like Pierre-Robin, Stickler, Weissenbacher-Zweymuller (W-Z), and Marshall syndromes are variable expressions of the same gene (Table 1). Due to analogous clinical manifestations, approximately half of the cases with Pierre-Robin anomaly have been suggested to have Stickler syndrome (Herrmann et al, 1975). The W-Z syndrome has been viewed as a neonatal expression of Stickler syndrome (Kelly et al, 1982; Winter et al, 1983) and has been included in the description of OSMED by Giedion et al (1982). The main manifestations of Stickler syndrome, an autosomal dominant disorder, are high myopia with retinal detachment and cataracts, flat nasal bridge, arthropathy with a mild spondyloepiphyseal dysplasia, cleft palate, bifid uvula, and hearing loss (Jacobson et al, 1990). Although the degree and nature of hearing loss in Stickler syndrome varies, approximately 50 percent of reported patients exhibit conductive hearing loss secondary to palatal abnormalities (Williams et al, 1981; Gould, 1989). Furthermore, a small percentage of patients with Stickler syndrome show anomalies of the malleus and incus (Igarashi et al, 1976). The prevalence of sensorineural hearing loss has been reported to be between 10 percent (Jung, 1989) and 87 percent (Liberfarb and Hirose, 1982) and may be due to cochlear malformation (Igarashi et al, 1976). Designation of individuals with severe midface hypoplasia as having Marshall syndrome has been debated. Some investigators suggest that the two conditions are the result of the same mutant gene (Cohen, 1974; Baraitser, 1982). The distinctive feature of OSMED is large epiphyses; in infancy, dumbbell-shaped deformity of the short femur is typical, whereas moderate platyspondylia, large epiphyses with pathognomonic appearance of hands, occurs in early childhood (Giedion et al, 1982).

This paper describes two siblings identified with OSMED at the age of 3 years. These cases presented with classic features of OSMED including sensorineural hearing loss, short extremities with normal body length, and abnormally thick joints. The siblings also presented with high myopia and a pattern of inheritance often associated with Stickler-Marshall syndrome.

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Table 1 Characteristic Features of Some Skeletal Dysplasias Compared with Present Case Studies

Features	*Marshall*	*W-Z*	*Robin*	*Stickler*	*OSMED*	*Case Studies*
Hearing loss						
Conductive*	—	—	+	+	—	—
Sensorineural	+	+	—	+	+	+
Inheritance						
Autosomal recessive	—	—	—	—	+†	—
Autosomal dominant	+	+†	+‡	+‡	—	+
Cleft palate or bifid uvula	+	+	+	+	+	—
Severe myopia	+	+	+	+	—	+
Midface hypoplasia	+	+	+	+	+	+
Micrognathia	+	+	+	+	+	—
Large epiphyses	—	+§	—	—	+	+

*Conductive hearing loss may be associated with palatal abnormalities.
†Giedion et al (1982) suggested that OSMED includes W-Z.
‡Jacobson et al (1990) differentiated Stickler and Robin in the pattern of inheritance.
§Cortina et al (1977) indicated small epiphyses.

CASE REPORTS

Case history revealed several family members with hearing, vision, and skeletal problems (Fig. 1). The great grandmother of the siblings (generation I, Fig. 1) and three of her four children (generation II, Fig. 1) had all of the above manifestations. These three affected children each had at least one offspring who displayed hearing, vision, and skeletal problems. The normal fourth child had two offspring who were not affected (generation III, Fig. 1). This data suggests a dominant pattern of inheritance.

Ten family members (generations II through IV, including the described case studies) received amplification and/or aural rehabilitation services at the Cleveland State University clinic. Review of the audiologic findings of these family members indicated a bilateral sloping sensorineural hearing loss varying from mild to severe degree. All reported family members were oral/aural and several were fitted with amplification initially as adults. Case history information suggested that the hearing loss of these members remained essentially unchanged since the initial diagnosis. Since there was no evidence of palatal abnormalities (cleft palate or bifid uvula) in any family member, conductive hearing loss was limited to the occurrence of transient middle ear problems.

Case 1

Sibling one is a 14-year-old female functioning successfully in a ninth grade mainstream classroom. Audiometric test results showed a sloping mild to severe sensorineural hearing loss in both ears (Tables 2 and 3, Figure 2A). Aided warble tone thresholds indicated 20 to 30 dB of gain using a Phonak Audinet PPC-C ear level hearing aid fitted for the right ear (SSPL: 125, low cut: H, vol: 2 coupled to a soft shell mold). Results of aided speech audiometry showed satisfactory word recognition scores at soft conversational level (84% at 40 dB HL). Measurement of auditory evoked potentials at

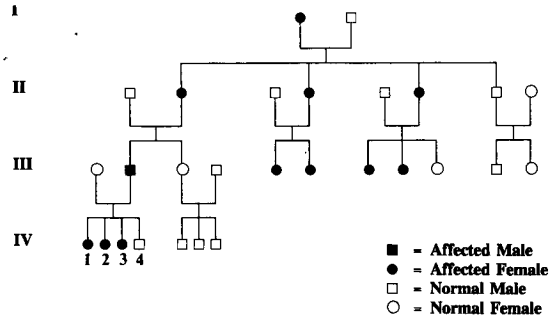


Figure 1 Family tree as reported by the mother of OSMED cases (2 and 3).

Table 2 Results of Speech Audiometry for Sibling One

Ear	*SRT (dB HL)*	*Word Recognition* (%/dB HL)*
Right	40	60/80
Left	30	76/70
Aided†	20	84/40

* Performed live voice with CID W-1 spondees and NU #6 monosyllabic word list.
†Aided monaurally (Phonak Audinet); SSPL: 125; low cut: H; vol: 2; soft shell mold.

Table 3 Results of Acoustic Reflex Measurements for Sibling One

	Right Ipsilateral				*Right Contralateral*			
Frequency (Hz)	500	1000	2000	4000	500	1000	2000	4000
Threshold (dB HL)	95	110	NR	NR	90	90	95	100
Decay	None	None	—	—	None	None	—	—
	Left Ipsilateral				*Left Contralateral*			
Threshold (dB HL)	80	80	85	85	95	105	110	115
Decay	None	None	—	—	None	None	—	—

NR = no response.

high intensity levels showed normal brainstem, middle latency, and late responses (Fig. 2B).

Based on initial assessments (at age 9 years, 11 months), aural rehabilitation focused on expressive language, especially in the areas of syntax and morphology, development of auditory comprehension skills in quiet and background noise, and articulation therapy for improvement in intelligibility of connected speech. Current assessments indicate age-appropriate receptive and expressive language abilities (total language score: 100; reference score: 100 on Clinical Evaluation of Language Fundamentals-Revised [CELF-R; Semel et al, 1987]) and successful completion of subtests 9 and 10 of the Test of Auditory Comprehension, which evaluate the ability to sequence three events and recall five details in the presence of competing messages (T scores: 51 and 59, respectively; reference T score: 50 [TAC; Trammell et al, 1977]). These achievements are remarkable for the degree of hearing loss, especially in light of the visual limitations (prosthetic left eye due to retinal detachment and high myopia 20/50 in the right eye).

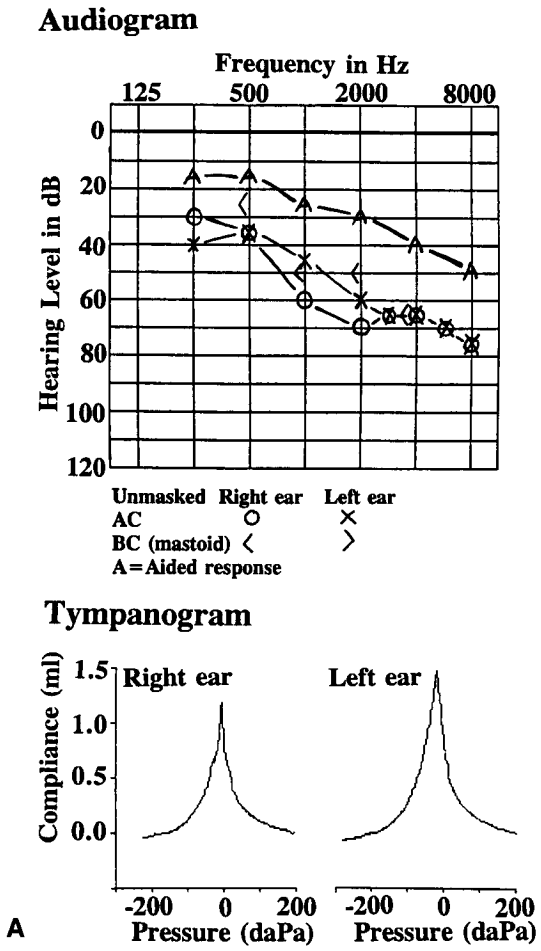
Case 2

Sibling two is an 11-year-old female who is academically challenged in a fifth grade mainstream classroom. Audiometric results indicated moderately severe to profound sensorineural hearing loss in both ears (Table 3 and Fig. 3). Aided warble tone thresholds revealed a gain of approximately 50 dB using binaural Phonak Super Front PP-C4 ear level hearing aids (SSPL: 5, low cut: 1, high cut: 0, vol: 3.5 coupled to soft shell earmolds). Aided word recognition tests indicated mildly reduced scores at high presentation levels.

Due to the severity of hearing loss, sibling two was enrolled in an auditory-oral preschool program for hearing-impaired children at the age of 4 years. During kindergarten and first grade, she was mainstreamed half time and was fully integrated in a regular classroom by second grade. Aural rehabilitation therapy was initiated at the age of 7 years for development of auditory skills in the areas of discrimination, memory, and comprehension. Delays in articulation, morphology, and syntax were also addressed. The most recent evaluations showed fair to poor receptive and expressive language abilities (total language score: 59; reference score: 100 on CELF-R). These language test results explain the difficulties that sibling two is experiencing in reading comprehension and writing. Her receptive abilities are further compounded by her severe vision problems, diagnosed as high myopia in both eyes (visual acuity in the right eye: 20/70 and the left eye: 20/200). Results of the Test of Auditory Comprehension showed ability to sequence three events under quiet conditions and recall five details with competing messages (T scores: 61 and 76, respectively; reference T score: 50). These auditory skills are above expected levels for her age group and severity of hearing loss.

COMMENT

Although the hearing loss of both siblings was identified at the age of 3 years, appropriate intervention including amplification and aural rehabilitation permitted early educational mainstreaming. Sibling one demonstrated excellent cognitive/auditory and language abilities explaining her outstanding performance in the classroom. Aural rehabilitation therapy is currently focused on articulation remediation in connected speech. Sibling two has intelligible speech and strong cognitive/auditory skills under quiet conditions, but significant deficits in receptive



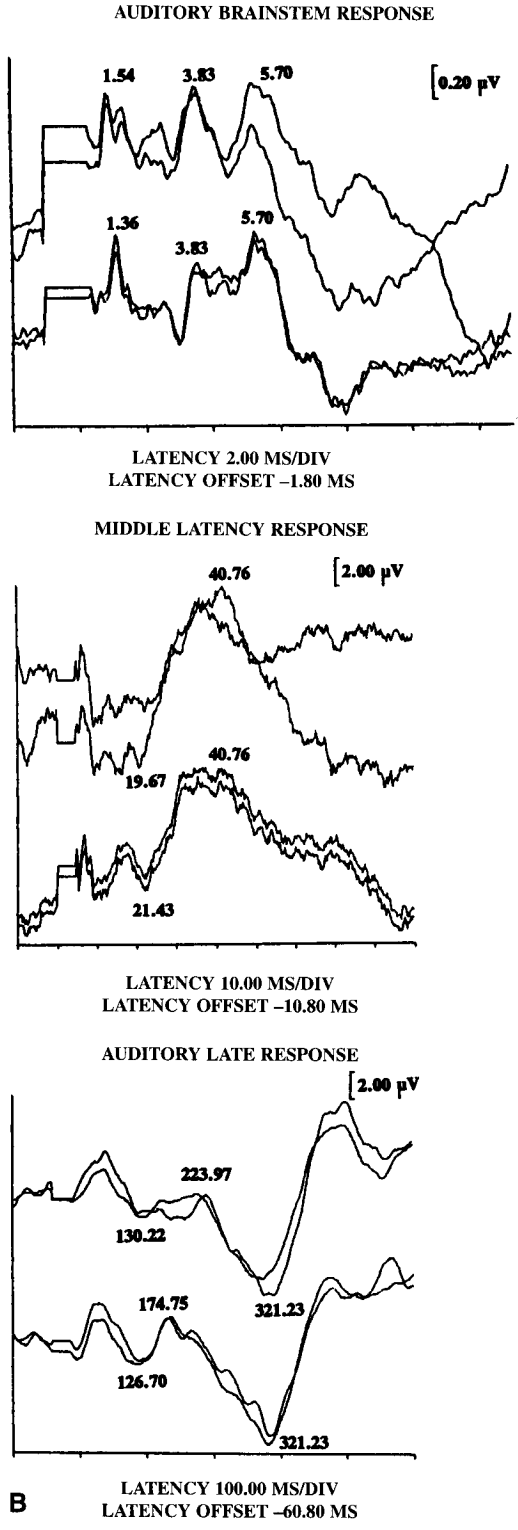


Figure 2 *A*, Audiologic test results for sibling one; *B*, normal auditory brainstem and middle latency responses to clicks and normal late responses to 250 Hz tone bursts (Fz-A1/A2) delivered at 70–95 dB nHL are also shown.

and expressive language are affecting her classroom performance. Given the listening environment and complexity of oral information presented in the mainstream classroom, an assistive listening device may greatly enhance her educational performance.

Both cases presented in this study showed severe myopia, a feature not seen in OSMED cases. Further, the dominant mode of inheritance indicated by the family tree (see Fig. 1) is consistent with that of Stickler-Marshall syndrome and in contrast with the recessive pattern

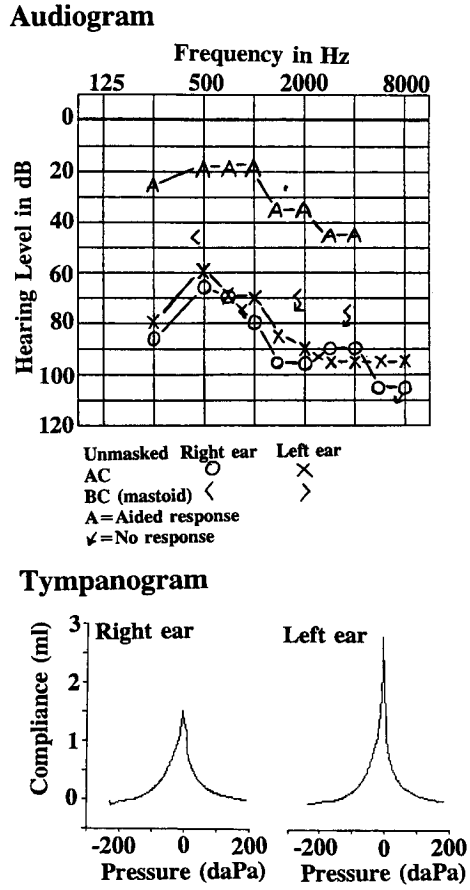


Figure 3 Audiologic test results for sibling two.

suggested by Giedion et al (1982). However, presence of large epiphyses in these cases is a characteristic feature of OSMED, compared to the small epiphyses documented in Stickler-Marshall syndrome. Thus, the siblings described in this study show features of both OSMED and Stickler-Marshall syndrome. Such combinations of findings support the postulation that OSMED and some skeletal dysplasias are variable expressions of the same gene. Regardless of differential diagnosis, since one of the manifestations of syndromes associated with skeletal dysplasias is hearing loss, early identification is critical.

Table 4 Results of Speech Audiometry* for Sibling Two

Ear	*SRT (dB HL)*	*Word Recognition† (%/dB HL)*
Right	65	92/95
Left	60	81/90
Aided‡	45	80/65

*Acoustic reflexes could not be elicited at the maximum limits of the equipment for all of the test conditions (right/left ipsilateral; right/left contralateral) across all frequencies.

†Performed live voice with CID W-1 spondees and WIPI word list.

‡Aided binaurally (Phonak Super Front); SSPL: 5; low cut: 1; high cut: 0; vol: 3.5; soft shell molds.

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