X-Linked Mixed Hearing Loss with Stapes Fixation: Case Reports

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Abstract

X-linked mixed hearing loss with stapes fixation is a congenital, recessive anomaly. The condition is characterized by progressive mixed hearing loss, reduced vestibular responses, and congenital fixation of the stapes with perilymph gusher encountered during surgical intervention. This paper describes the audiologic history of a family with two affected male children and one carrier female child. The need for early intervention and the challenges encountered in evaluation and management are discussed.

Key Words: Amplification, congenital stapes fixation, perilymph gusher, X-linked mixed hearing loss

X-linked (sex-linked) recessive forms of deafness are rare, accounting for less than 3 percent of all congenital deafness (Fraser, 1976). Most cases of congenital deafness are sensorineural in nature; however, an X-linked recessive form of mixed hearing loss also exists. This discussion focuses on members of a family exhibiting X-linked mixed hearing loss with stapes fixation and perilymph gusher at stapedectomy. Of the three children in this family, both male children are affected, and the female child is a carrier. These cases highlight the importance of early intervention, the challenges in audiometric testing, and the necessity for frequent follow-up in patient management.

BACKGROUND

Hereditary mixed hearing loss with stapes fixation and perilymph gusher was initially inferred by Olsen and Lehman (1968) and reported as an X-linked inheritance by Nance et al (1971). Other cases have subsequently been reported (Glasscock, 1973; Jensen et al, 1977; Cremers et al, 1983, 1985; Bento and Miniti, 1985; Phelps et al, 1991). A congenital, recessive anomaly, it has been connected to a defective gene that lies somewhere in the q13–21 region along the long arm of the X chromosome (Bruner et al, 1988; Wallia et al, 1988).

X-linked recessive traits affect males through female links. The mothers and daughters of affected males are obligate carriers. A carrier female will pass the defective gene on to 50 percent of her sons and 50 percent of her daughters. The X-linked trait is unique in that there is no direct male-to-male transmission. This particular X-linked condition is characterized by a bilateral, progressive mixed hearing loss, reduced vestibular responses, congenital fixation of the stapes, and a perilymph gusher when a stapedectomy is attempted. Affected individuals do not always develop functional speech and language skills.

Anatomic findings of an abnormal dilatation of the lateral extent of the internal auditory canal have been confirmed through polytomography (Glasscock, 1973; Bento and Miniti, 1985; Cremers et al, 1985) and high resolution computed tomography (CT) scan (Phelps et al, 1991). Additionally, a deficiency or absence of bone between the lateral portion of the internal auditory meatus and the basal turn of the cochlea (Bento and Miniti, 1985; Cremers et al, 1985; Phelps et al, 1991) and cochlear malformations have also been reported (Bento and Miniti, 1985; Cremers et al, 1985; Phelps et al, 1991).
MANIFESTATIONS OF HEARING LOSS

The typical audiometric finding for affected males is a bilateral, symmetrical, severe-to-profound mixed hearing loss. The loss shows a progression in the sensorineural component (Cremers et al., 1985), which may demonstrate a fairly rapid onset (Glasscock, 1973). The conductive component is greatest in the low frequencies (Cremers et al., 1985), consistent with the expectations of a stiffness pathology. Affected individuals require some form of educational assistance and are frequently placed in classrooms for the hearing impaired (Cremers et al., 1985). Speech and language skills resemble those of an individual with a profound sensorineural hearing loss. Female carriers have been reported to show a mild-to-moderate mixed or sensorineural hearing loss (Nance et al., 1971; Cremers and Huygen, 1983; Phelps et al., 1991), but more often display normal hearing sensitivity.

The cases presented are three children, two male and one female, who have been followed in The University of Texas Medical Branch audiology clinics for a period of 10 years. The affected male children have different fathers. A description of our experiences in evaluating and habilitating these children is presented.

CASE STUDIES

Case 1

MB was initially referred to the audiology clinic at 2 years of age (September, 1982). The referral was initiated as a routine part of a developmental evaluation through the child development clinic. He presented to our clinic with delayed speech and language development, a familial history of hearing loss, and familial megencephaly. Pressure equalization (PE) tubes had been placed bilaterally by his pediatrician in March of 1982. Sound-field audiometric testing was consistent with a severe hearing loss for the better ear, the nature of which could not be determined at that time (see Fig. 1). PE tubes were found to be nonpatent, consistent with the possibility of a conductive component to the hearing loss.

Testing 1 month later revealed sound-field responses averaging 65 dB HL, agreeing with those previously obtained. Immittance testing (tubes out) showed normal middle ear mobility (type A tympanograms) and absent acoustic reflexes. Since sound-field responses were unchanged from the previous evaluation, and the tympanogram showed normal mobility, the sensitivity loss was believed to be sensorineural in nature. An auditory brainstem response (ABR) threshold was obtained on the same day at 75 dB nHL for the right ear, adding credibility to the results of the sound-field testing. ABR data were not available for the left ear.

Medical examination, tomograms, and all lab tests were normal. A pedigree of four hearing-impaired males in four generations, however, was revealed through genetic counseling. Since all were related through females, the pattern of inheritance was considered to be compatible with an X-linked, recessive sensorineural hearing loss. As a carrier of the gene, MB's mother, who had normal hearing, was counseled that a recurrence risk of 50 percent existed for passing the gene on to either a male or female child. She was informed of a one in four risk for having a hard-of-hearing son.

An educational program and amplification were recommended in October of 1982. The family made plans to carry out these recommendations through a clinic in the vicinity of their home. Amplification was fitted in January of 1983.

In December of 1982, MB returned to our clinic with chronic otitis media. Tube placement occurred for the second time in February of 1983. Behavioral sound-field audiometric testing in March again suggested a severe hearing loss. At that time (2 years, 6 months of age), MB was successfully conditioned for unmasked bone-
conduction testing, which revealed the presence of a conductive component for at least one ear (see Fig. 2). Aided thresholds were recorded at levels of 30 to 40 dB HL in the 500 to 2000 Hz range. An aided speech-awareness threshold was also obtained at 35 dB HL.

MB was re-evaluated 1 year later (March, 1984). Air- and bone-conduction thresholds obtained under earphones were consistent with those previously obtained. A CT scan revealed an angulation and widening of the internal auditory canal, which was attributed to either a normal variant or the possibility of neurofibromatosis (though it was noted that clinical signs were not present at that time).

MB was followed for acute ear infections throughout the next year, with a third set of PE tubes placed in March of 1985. Threshold ABR testing under general anesthesia again revealed 70- to 80-dB nHL thresholds to air-conducted clicks and 30-dB nHL thresholds to bone-conducted clicks for each ear. A CT scan obtained following surgery was considered normal. Audiometric follow-up continued at regular intervals throughout the next 3 years and showed an essentially stable severe-to-profound mixed hearing loss bilaterally (see Fig. 3). Tympanometry revealed type A tympanograms and absent acoustic reflexes throughout these test intervals.

At 6 years of age, MB underwent surgery for a right stapedotomy. Exploration of the middle ear space revealed the presence of adhesions between the incus, eardrum, and promontory, which were subsequently separated. The malleus and incus were mobile, but the stapes was found to be fixed. The surgery was interrupted when a perilymph gusher occurred at perforation of the footplate. Following control of the gusher, a de la Cruz piston was placed and the procedure concluded.

Postoperative audiometric testing revealed a profound hearing loss in the right ear. Masked bone-conduction testing could not be carried out, due to the degree of hearing loss. A change in test behavior and history, however, supported the assumption of an increased sensorineural component. Postoperatively MB showed different reactions to stimuli presented to the right ear. He demonstrated increased distractibility and other behaviors (e.g., pressing on earphones, confusion, displeasure) during testing of the right ear, which were not present preoperatively or while testing the left ear.

MB continues to be followed and has a stable, profound hearing loss in the right ear and a profound-to-severe mixed hearing loss in the left ear (see Fig. 4). He was first aided at 2 years of age and currently wears binaural, high-gain, behind-the-ear (BTE) hearing aids. Since his surgery, however, he generally shows preference for the left aid. Bone-conduction aids have been used for speech training during times of chronic drainage and during problems with the earmold fit following stapes surgery. Functional gain measures suggest aided responses in the 30- to 40-dB HL range.

Speech and language therapy have been provided by the local regional deaf education program since MB's enrollment at the age of 2 (initial diagnosis). A total communication program has been emphasized. His speech and language skills have not followed the "normal" pattern of development expected with a largely conductive hearing loss. Instead, MB has demonstrated speech-language characteristics similar to that expected with a profound sensorineural hearing loss. MB was diagnosed as having attention deficit disorder in 1988 and subsequently placed on Ritalin for his hyperactivity. He continues to be followed in the child development clinic for his behavioral and speech/language difficulties.

Case 2

KM is the half brother of MB and was delivered by repeat cesarean section in July of 1987. He was a full-term birth and had a normal...
birth weight (8 lbs, 8 oz). KM had a positive family history of Charcot-Marie-Tooth disease that occurred in every other male and one female on the paternal side. Two days following birth, he was treated with antibiotics for sepsis and remained hospitalized for 10 days. ABR screening (air-conduction click) at 2 days of age revealed responses at 60 dB nHL for each ear. Bone-conduction ABR responses were questionable at 30 dB nHL. ABR testing at 1 month of age yielded similar results. KM was immediately enrolled in a home-based infant stimulation program.

Behavioral observation audiometry at 5 months of age could not rule out a moderate hearing loss. Immittance audiometry revealed type B tympanograms, supporting a conductive component. Binaural moderate gain amplification was fitted 1 month later. KM currently exhibits a severe mixed hearing loss (see Fig. 5) with fluctuations secondary to chronic middle ear problems. Surgical intervention has not been attempted, based on the expectation of perilymph gusher at surgery.

KM has had six sets of PE tubes over the past 5 years. He currently wears binaural, moderate-gain, behind-the-ear hearing aids at home. His use of amplification has been inconsistent over the past 4 years as a result of chronic otitis media, otorrhea, and upper respiratory congestion (related to food allergies). Bone-conduction amplification has been utilized to promote consistent amplification during these times. KM has shown a preference for an FM auditory training system, which he uses at school. We continue to monitor his performance with both the BTE aids and the FM device to determine the reason for this preference.

KM entered a school-based, deaf-education class at 3 years of age. He attends a full-day program emphasizing total communication. After school, he receives additional speech and occupational therapy at a rehabilitation center near his home. He continues to demonstrate severe developmental delays in speech-language, visual-motor integration, and fine motor skills. These difficulties contribute to his persistent insecurity and apprehension in the test situation.

Case 3

HM is the sister of KM and half-sister of MB. ABR screening at birth (September, 1988) revealed responses at 45 dB nHL for each ear. Risk factors included familial history of hearing loss and left postauricular skin tag. Behavioral sound-field and immittance testing conducted at 8 months of age revealed a mild-to-moderate hearing loss with probable conductive component. The ear tag was removed and PE tubes placed at that time, resulting in improved hearing threshold levels.

HM demonstrated a fluctuating conductive hearing loss secondary to chronic otitis media through 18 months of age. She has had two sets of PE tubes over the past 3.5 years but continues to show full recovery of hearing levels following treatment. Her current hearing levels are shown in Figure 6. Developmental milestones have been age appropriate. Audiometric milestones have been age appropriate. Audiometric follow-up continues on at least an annual basis because of the increased prevalence of hearing loss in carriers and siblings of individuals affected by this condition.
SUMMARY

An X-linked mixed hearing loss, though infrequent, can pose many challenges to the evaluation of hearing. Information gained in the test situation can be limited and may require further confirmation. As in the case of MB, assumptions concerning the nature of hearing loss should be avoided until responses to some form of bone-conduction testing can be obtained. Dependence on impedance results to infer the nature of a severe-to-profound hearing loss can be misleading. In MB's case, the results of a severe hearing loss with type A tympanograms led the audiologist to erroneously assume a sensorineural hearing loss. The absence of acoustic reflexes were believed to be consistent with the degree of hearing loss, without considering the possibility of a problem (e.g., fixation) within the middle ear system.

Additional challenges are created through the masking dilemmas imposed by the degree (severe to profound) and nature (large conductive component) of this hearing loss. Even when audiometrically possible, the use of masking is often restricted by the behavioral and developmental deficits exhibited by individuals with X-linked mixed hearing loss. Decreased attention span, distractibility, emotional hypersensitivity, and motor and speech/language delays limit the amount of information that can be obtained, particularly in the early stages of evaluation. Thus, it may be difficult to determine the true sensitivity and effects of the hearing loss. Definitive differentiation between pure sensorineural versus mixed hearing loss is important in the selection of gain for appropriate amplification and allows the audiologist to explore alternatives (e.g., bone-conduction aids) in hearing aid management. Recognition of the nature of the hearing loss is also important to guide the audiologist in making recommendations to educational personnel. For example, the individual with a mixed hearing loss would typically be expected to have better potential for using auditory cues than one with a sensorineural hearing loss of the same degree. Individuals with X-linked mixed hearing loss may, however, show communication behaviors similar to those of a profoundly hearing-impaired individual.

A careful family history serves to alert the clinician to the possibility of X-linked recessive hearing loss and the need for aggressive management. In the case of KM, knowledge of this hereditary condition led to a more aggressive approach to evaluation and management. An initial diagnosis of middle ear pathology in an infant would commonly be handled with medical management and frequent audiologic monitoring rather than pursuance of amplification and educational programming.

The progressive nature of this type of hearing loss necessitates frequent audiometric monitoring. As in the case of KM, awareness of the added fluctuations in hearing levels due to chronic middle ear problems is important. These temporary sensitivity changes must be differentiated from any progression of the loss and provide information essential for amplification management. Flexibility of amplification strategies is critical for maximizing auditory stimulation for the continued development of speech and language skills. Bone-conduction aids were
helpful for both MB and KM at points in time when traditional forms of amplification were medically contraindicated. The use of an FM system was also beneficial for KM.

Finally, audiologists should be aware of the problem of perilymph gusher (at surgical intervention) associated with this syndrome. The audiologist who recognizes the audiometric and behavioral characteristics associated with X-linked mixed hearing loss can contribute information important to the medical management process. Knowledge of the possibility of this syndrome and the associated problems may be useful to the physician when making decisions related to surgical intervention.

REFERENCES


