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Congenital Conductive Hearing Loss in the Lacrimoauriculodentodigital Syndrome

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An inherited middle ear anomaly that was causing hearing impairment in a 12-year-old girl was treated successfully by a stapedotomy combined with a malleovestibulopexy. Cup-shaped ears, abnormal or absent thumbs, and skeletal deformities of the forearms were present in several members of 3 generations of a family. An autosomal dominant pattern of inheritance was recognized. These features are present in a number of previously described syndromes, but they correspond best with the lacrimoauriculodentodigital syndrome.

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In about one third of patients with congenital conductive hearing loss, the hearing loss is part of a syndrome. About 50 genetic syndromes with congenital conductive hearing loss as a feature have been reported. The lacrimoauriculodentodigital (LADD) syndrome is new to this list. LADD syndrome is listed as a separate entity in the catalogs by McKusick,¹ and an autosomal dominant pattern of inheritance was recognized. In most cases of LADD syndrome, the hearing loss is pure sensorineural. In the case reported herein, the hearing loss is congenital in origin and of the conductive type. Reconstructive surgery of the ossicular chain proved to be successful.

REPORT OF A CASE

A 12-year-old girl (**Figure 1**) was referred for reconstructive middle ear surgery. Exploratory tympanotomy had been performed on the left side at another institution when the patient was 5 years old. At that time, a dysplastic incus body was removed; there was no stapes superstructure, and the stapes footplate was ankylosed. Bilateral dysplasia of the superior helixes was corrected at that time.

Otologic examination showed normal tympanic membranes. The auricles were smaller than normal (**Figure 2**). Audiometry indicated flat air conduction thresholds of 60 dB in the left ear and 20 dB in the right ear, with absent ipsilateral and contralateral stapedial reflexes (**Figure 3**). Middle ear compliance and pressure were normal. Computed tomographic scanning of the petrosal bones demonstrated normal inner ear structures.

Interdentate spacing in the upper and lower arches was abnormal. The small permanent incisors had the appearance of deciduous teeth. Although the enamel of all the teeth was thin, premature decay had not occurred. No third molar teeth were present. The hard and soft palates were normally fused, with no clefts.

The left thumb was hypoplastic and digitlike; flexion was restricted. The left thenar eminence was hypoplastic (**Figure 4**). Radiographs of the hands and feet were normal. Results of ophthalmologic examination were also normal; dacryostenosis was absent, and all lacrimal puncta were present.

During explorative tympanotomy of the left ear at the age of 12 years, slight fixation of the head of the malleus was noted in the epitympanum. The incus had been removed during the previously performed tympanotomy. Stapes footplate ankylosis was confirmed. The malleus head

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Figure 1. A 12-year-old girl with the lacrimoauriculodentodigital syndrome and unilateral congenital conductive hearing loss.

was removed and the tympanic membrane was stripped from the proximal one third of the long process of the malleus. Stapedotomy was performed on the footplate of the stapes, and a 0.4-mm Teflon-platinum piston (Richards Co, Memphis, Tenn) with a length of 4.75 mm was used to perform a malleovestibulopexy. The piston eye was fixed around the proximal part of the malleus handle, which had been partly stripped of the tympanic membrane.

Postoperative results indicated improvement in the conductive threshold from 60 dB preoperatively to 20 dB postoperatively, with a 5-year follow-up (Figure 3).

A number of members of the proband's family had similar characteristics. The proband's mother had a left-sided facial asymmetry. Both helixes had been corrected. Thumbs and fingers were normal, and function was unimpaired. Supination at the elbows was reduced. Age-related bilateral flat sensorineural hearing loss was present. The proband's sister was normal.

Investigation of a maternal uncle demonstrated dysplasia of both auricles, slight hypoplasia of the left shoulder musculature, and hypoplasia of both forearms and hands that was more severe on the left side. The left thumb was missing, while the right thumb and index finger were hypoplastic. Radiographs of the forearms showed bilateral short forearm bones, more marked on the left, with hypoplasia of the olecranon,

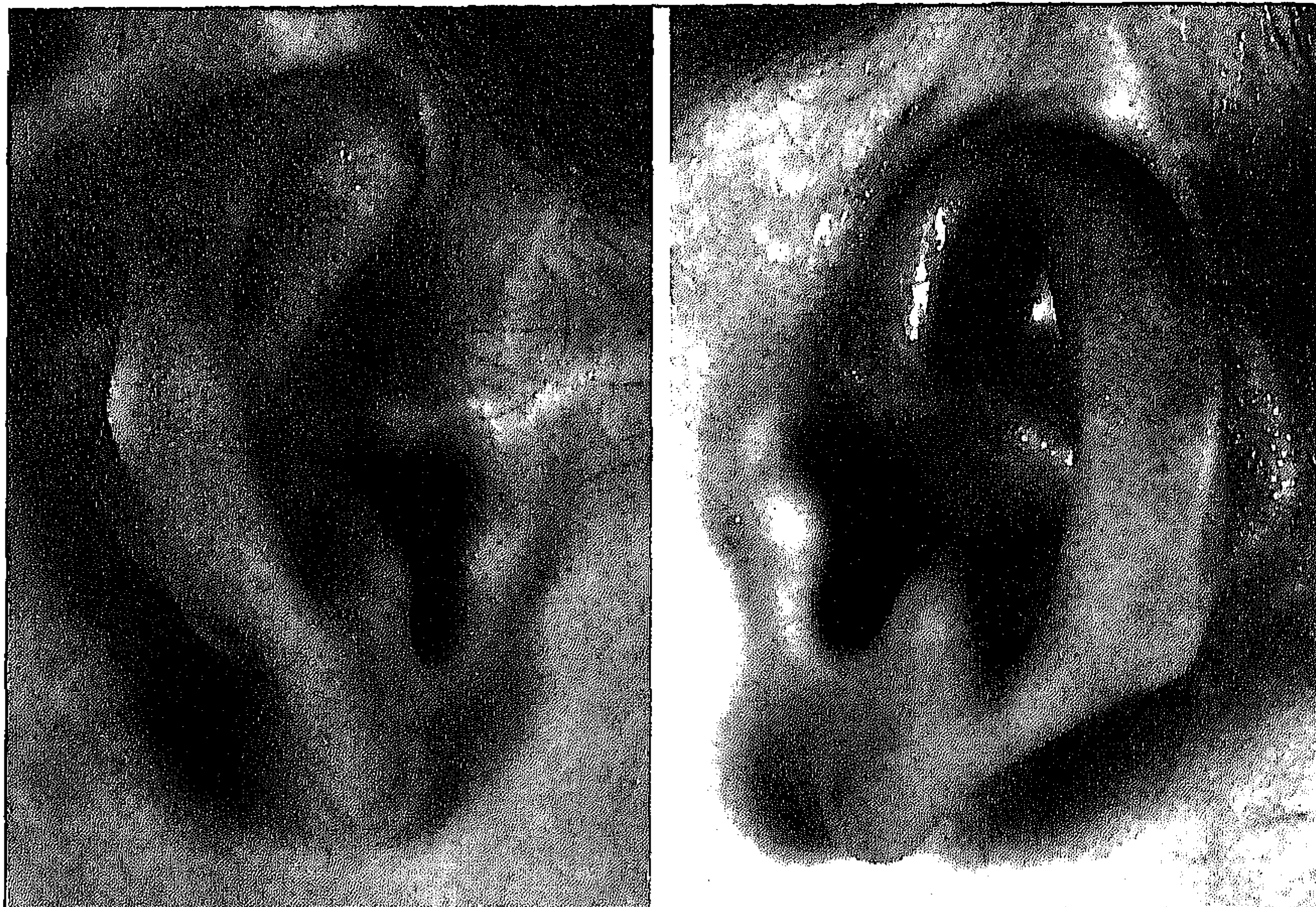


Figure 2. Auricles of the proband. Note the scars from auriculoplasty.

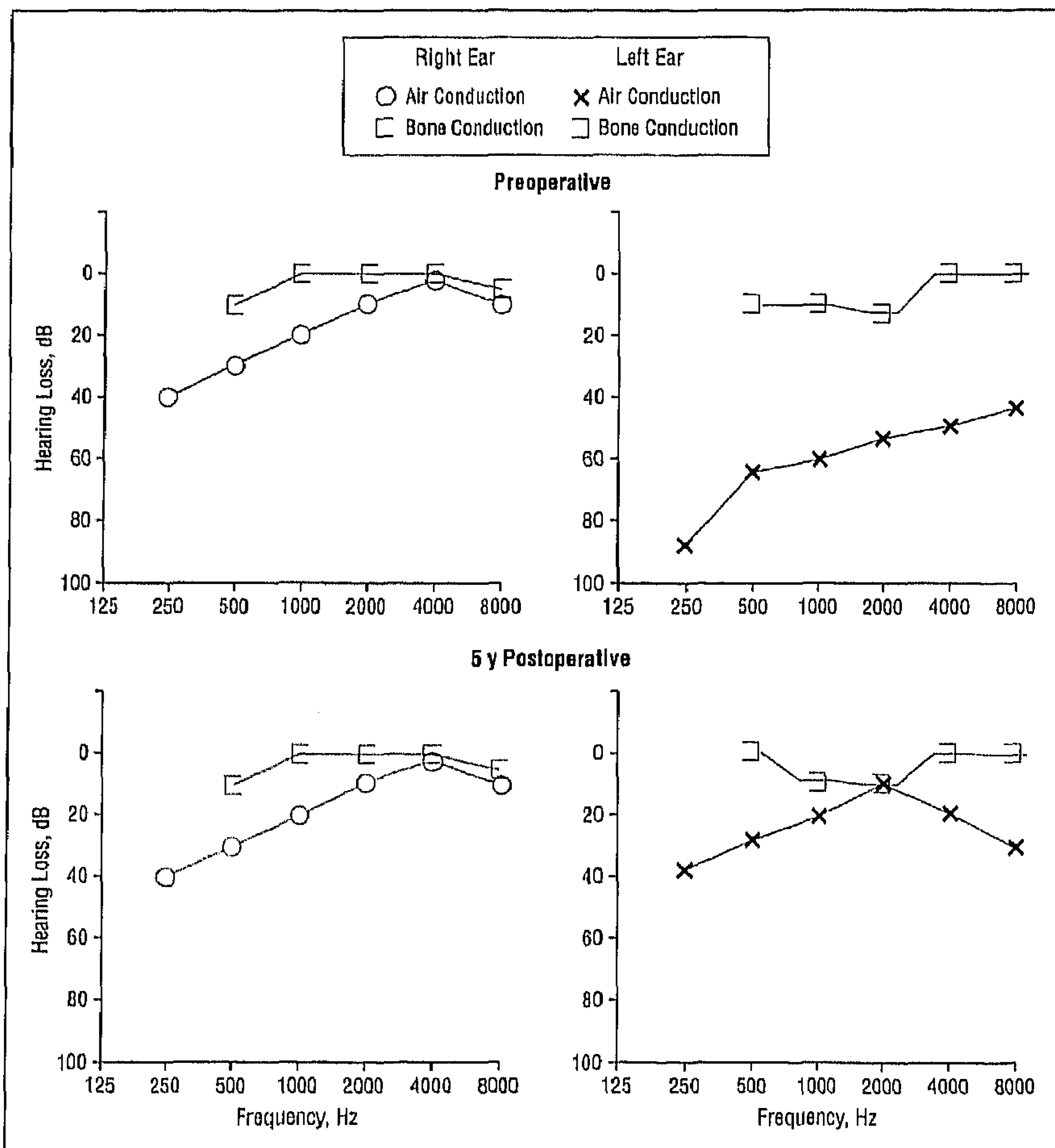


Figure 3. Pure tone audiograms obtained preoperatively (top) and 5 years after malleovestibulopexy (bottom).

coronoid process, ulna, and the head of the radius. Synostosis of the trapezium, trapezoid, capitate, and pisiform bones in both hands had occurred. Audiometric data were not

available, but his hearing was reported to be normal.

The proband's grandfather was reported to have had cup-shaped ears and had never been able to ex-

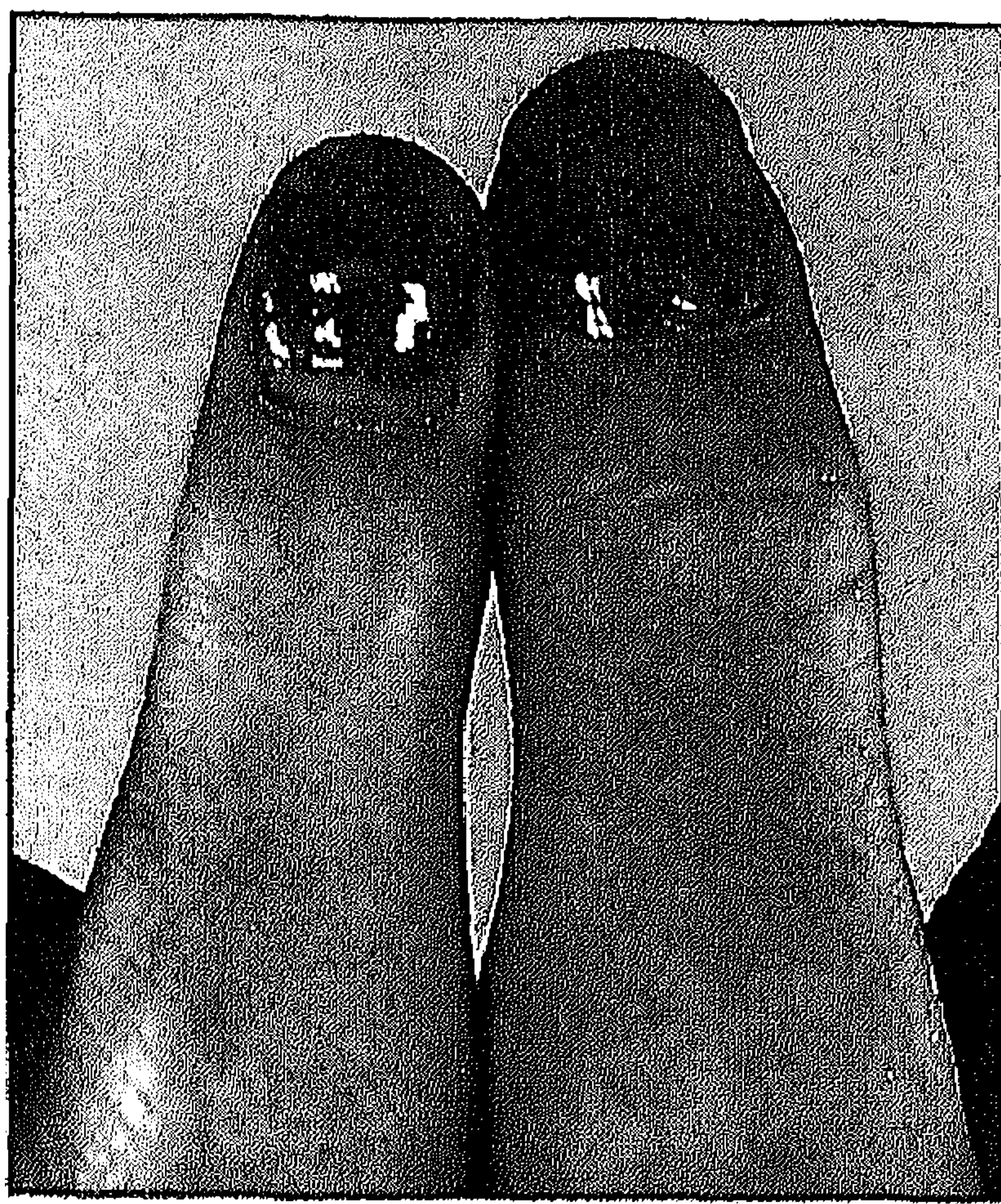


Figure 4. Anomalies of the thumbs, with a digitlike aspect of the left thumb.

tend his thumbs fully. Precise history about hearing function was not known.

COMMENT

The features observed in the present family correspond most closely to the report by Hollister et al² of 2 generations of a family who had cup-shaped ears, mixed hearing loss, and nasolacrimal duct obstruction, and to an isolated case described by Levy³ diagnosed as LADD syndrome. This syndrome is now listed as a separate entity in McKusick's catalogue.¹

Syndromes with an autosomal recessive pattern of inheritance, cup-shaped ears, and middle ear anomalies were not considered to apply to our patient.⁴⁻⁶ Syndromes with an autosomal dominant pattern of inheritance (eg, the branchio-otorenal syndrome) could also be excluded from the differential diagnosis because, although cup-shaped ears were present, there were no branchial fistulas and preauricular sinuses.⁷ In a similar syndrome, cup-shaped ears, commissural lip pits, and ear pits are present, but skeletal abnormalities have never been reported.⁸

The hand anomaly observed in the proband did not resemble ectrodactyly, and none of the subjects had a cleft lip and palate, as is present in the ectrodactyly-ectodermal dysplasia clefting syn-

drome.⁹ Another syndrome is characterized by radial ray aplasia, mixed or sensorineural hearing loss, external ophthalmoplegia, and thrombocytopenia. Skeletal abnormalities are more severe, and cup dysplasia is absent.¹⁰

After the first description of LADD syndrome, similar observations have been described by several authors.^{2,3,11-13} Limb malformations are the most common feature, but they are also highly variable. Teeth are often discolored, and enamel thinning occurs. Premature decay leads to the loss of permanent teeth, often before adolescence. Various salivary gland anomalies have also been described, leading to xerostomia, which enhances tooth decay. Cup-shaped ears are mentioned consistently and are usually more obvious than the hearing loss. Other features, such as weeping eyes caused by blockage of tear ducts, are also noticeable.

The incidence of hearing loss described in reports of the LADD syndrome is approximately 50%. In most cases, it consists of mild to severe predominantly sensorineural hearing loss.

This case of middle ear anomalies in association with the LADD syndrome was treated by reconstructive middle ear surgery. To perform a malleovestibulopexy, it is helpful to extend the upper part of the Rosen incision in an anterosuperior direction to achieve wider exposure of the malleus. The malleus head is then removed and the upper one third of the malleus is stripped of the tympanic membrane. The malleus handle is kept in position by the remaining anterior ligament. These procedures facilitate the crimping of the eye of a Teflon-platinum piston, although the malleus is in a much more anterior position than the normal anatomical position of the incus.

In congenital middle ear anomalies, the external auditory canal may be more curved than normal so that the anterior canal wall limits the surgical approach to the malleus. Reconstruction of the middle ear chain by a Teflon-

platinum malleus attachment prosthesis can help to overcome such anatomical limitations. Only a few reports of this procedure in cases of a missing incus and stapes arch have been published. Long-term results show closure of the pure tone average air-bone gap to within 10 dB in 70% of all individuals. Best results were reported in congenital chain defects.¹⁴ Our case, with a 5-year follow-up, illustrates a satisfactory outcome with this technique.

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Reprints not available from the author.

REFERENCES

1. McKusick VA. *Catalogue of Autosomal Dominant, Autosomal Recessive and X-Linked Phenotypes*. 10th ed. Baltimore, Md: Johns Hopkins Press; 1991:644-645.
2. Hollister DW, Klein SH, De Jager HJ, Lachmann RS, Rimoin DL. The lacrimo-auriculo-dento-digital syndrome. *J Paediatr*. 1973;83:438-444.
3. Levy WJ. Mesoectodermal dysplasia: a new combination of abnormalities. *Am J Ophthalmol*. 1969; 63:978-982.
4. Ellwood LC, Winter ST, Dar H. Familial microtia with meatal atresia in two sibships. *J Med Genet*. 1968;5:289-291.
5. Koningsmark BW, Nager GT, Haskins HL. Recessive microtia, meatal atresia and hearing loss. *Arch Otolaryngol Head Neck Surg*. 1972;96:105-109.
6. Mengel MC, Koningsmark BW, Berlin CI, McKusick VA. Conductive hearing loss and malformed low set ears as a possible recessive syndrome. *J Med Genet*. 1969;6:14-21.
7. Cremers CWRJ, Flikkers-van Noord M. The ear pits deafness syndrome: clinical and genetic aspects. *Int J Pediatr Otorhinolaryngol*. 1980;2: 309-322.
8. Marres HAM, Cremers CWRJ. Congenital conductive or mixed deafness, pre-auricular sinus, external ear anomaly, commissural lip pits: an autosomal dominant inherited syndrome. *Ann Otol Rhinol Laryngol*. 1991;100:928-932.
9. Rodini ESO, Richieri-Costa A. EEC syndrome: report on 20 new patients, clinical and genetic considerations. *Am J Med Genet*. 1990;37:42-53.
10. Arias S, Penchaszadeh VB, Pinto-Clisternas J, Larrauri S. The IVIC syndrome: a new autosomal dominant complex pleiotropic syndrome with radial ray hypoplasia, hearing impairment, external ophthalmoplegia and thrombocytopenia. *Am J Med Genet*. 1980;6:25-59.
11. Shiang EL, Holmes LB. The lacrimo-auriculo-dento-digital syndrome. *Pediatrics*. 1977;59:927-930.
12. Wiedemann HR, Drechsler J. LADD syndrome: report of new cases and review of the clinical spectrum. *Eur J Pediatr*. 1986;144:579-582.
13. Thompson E, Pembry M, Graham J. Phenotypic variation in the LADD syndrome. *J Med Genet*. 1985;22:382-385.
14. Tange RA. Ossicular reconstruction in cases of absent or inadequate incus, congenital malformations of the middle ear and epitympanic fixation of the incus and malleus. *ORL J Otorhinolaryngol Relat Spec*. 1996;58:143-146.