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Novel syndrome with conductive hearing loss and congenital glaucoma in three generations

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ABSTRACT

The objective of this paper was to describe the clinical and otological findings in multiple members of a family with congenital glaucoma, cardiac anomaly, and conductive hearing loss due to ossicular chain anomalies. We performed a retrospective review of the medical charts and otological materials of multiple members of the same family. Congenital glaucoma and hearing loss were inherited by the proband and her daughter, son, and mother, suggesting autosomal dominant inheritance. The son and daughter also showed atrial septal defects. Exploratory tympanotomies revealed anomalies of the long process of the incus in the proband and her daughter, and tympanoplasty improved hearing loss in both patients. This represents the first description of coexisting congenital glaucoma and conductive hearing loss due to ossicular chain anomalies in multiple members of a single family.

1. Introduction

Hereditary hearing loss is sometimes accompanied by ophthalmologic diseases such as Alstrom syndrome, Refsum syndrome, and Usher syndrome. Retinitis pigmentosa is related to these syndromes, and the affected individuals experience sensorineural hearing loss in most of these syndromes.

Familial conductive hearing loss caused by ossicular anomalies is rare. However, absence of the long process of the incus has been observed in multiple members of certain families. Higashi et al. [1] reported the cases of a mother and daughter with congenital conductive hearing loss caused by hypoplasia of the long crus of the incus. Kidowaki et al. [2] described cases of identical twins with congenital incudostapedial disconnection. Exploratory tympanotomies revealed absence of the long process but presence of the lenticular process of the incus in both ears of each twin. Nakanishi et al. [3] reported identical ossicular anomalies isolated to a mother and daughter, showing complete absence of the long process of the incus and fixation of the stapes. Wehrs [4] reported three generations of a family suffering from bilateral conductive hearing loss due to congenital absence of the long process of the incus. These reports suggest that absence of the long process of incus may be inheritable as either an autosomal dominant or X-linked dominant trait.

Hereditary hearing loss accompanying congenital glaucoma is very rare. We report herein the case of a family with congenital glaucoma, hypertelorism, and hearing loss, in which the cause of hearing loss in two members of the family was confirmed by exploratory tympanotomy.

2. Cases

The family pedigree is shown in Fig. 1. The proband and her mother showed hearing loss and congenital glaucoma. Both the daughter and son of the proband had hearing loss, congenital glaucoma, and atrial septal defect. The proband and her

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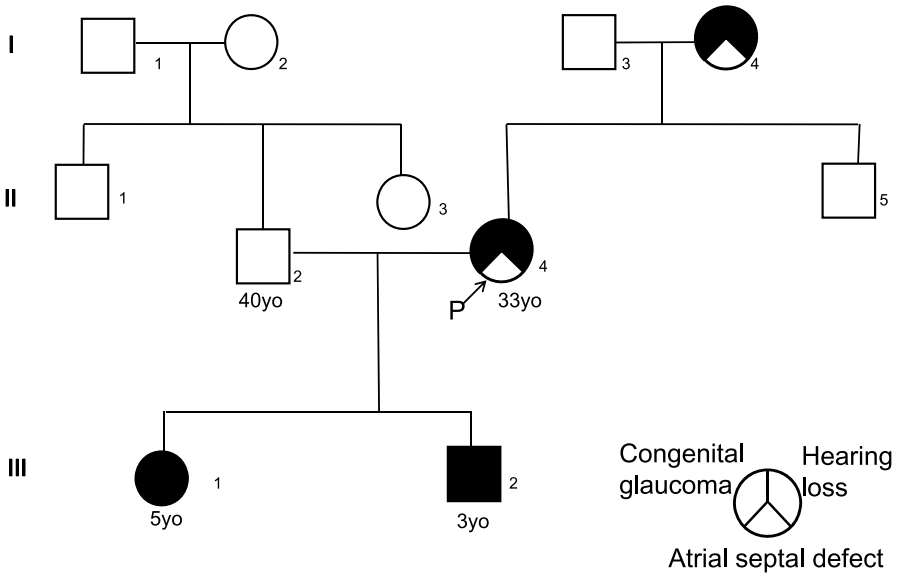

Fig. 1. Family pedigree of the family. The proband and her mother had hearing loss and congenital glaucoma. Both the daughter and son of the proband showed hearing loss, congenital glaucoma, and atrial septal defect.

children all had hypertelorism and had distinctive facial features (a flattened mid-face with a broad, flat nasal bridge, and a prominent forehead). Play audiometry of the son (III-2) showed bilateral conductive hearing loss (Fig. 2A). We offered genetic testing, but consent was not obtained.

Subject II-4 involved a 33-year-old woman who presented with a 1-year history of bilateral hearing loss. She had undergone surgery for congenital glaucoma at 2 months old. Bilateral eardrums were thin and pure tone audiometry showed bilateral conductive hearing loss with average hearing levels of

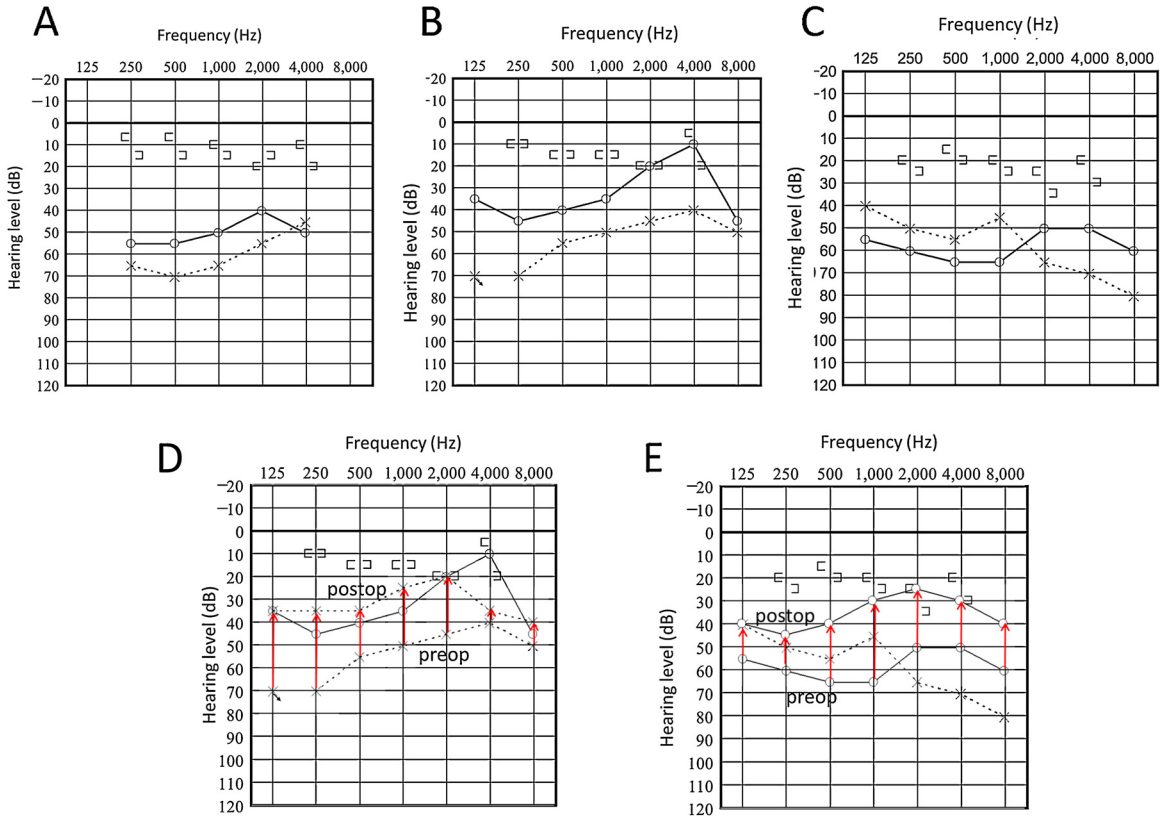

Fig. 2. Play audiometry of the proband's son (III-2) showing bilateral conductive hearing loss. (A) Preoperative pure tone audiogram of mother (B) and her daughter (C). Both pure tone audiometry showed conductive hearing loss, and hearing levels were higher in the daughter than in the mother. Pure tone audiometry performed about one year after the operation revealed improved hearing, with air conduction threshold of 26.3 dB (50 dB preoperatively) in mother (D) and with air conduction threshold of 31.7 dB (60 dB preoperatively) in her daughter (E).

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