Progressive Mixed-type Hearing Loss in an Adult Male with Osteopetrosis

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Introduction

- **Osteopetrosis** encompasses three distinct inherited diseases characterized by sclerotic bone lesions.
- Lesions result as a failure of normal osteoclast-mediated bone resorption.
- The incidence of osteopetrosis is estimated at 1 case per 100,000-500,000 live births.

- **Osteopetrosis congenita**
  - Autosomal Recessive
  - Presents in infancy
  - Characterized by: severe bone marrow failure, pancytopenia, bleeding, infection, failure to thrive, growth retardation, proptosis, blindness, deafness, hydrocephalus
  - Very poor prognosis

- **Marble Bone Disease**
  - Autosomal Recessive
  - Presents in childhood
  - Characterized by: no bone marrow failure, renal tubular acidosis, intracranial calcifications, hearing loss, psychomotor retardation
  - Poor prognosis

- **Osteopetrosis tarda**
  - Autosomal Dominant
  - Presents in adulthood
  - Characterized by: no bone marrow failure, brittle bones, increased susceptibility to fractures but with normal healing, hearing loss, degenerative joint disease, 50% of patients are asymptomatic
  - Good prognosis

Literature Review

- A Medline search for “Osteopetrosis” and “Hearing Loss” yields only 28 journal articles. The majority of these are case reports and/or from the general medical or radiological literature with scarce mentions of the otologic consequences of the disease.

- Notably:
  - There are two case series in the Otolaryngology literature, but both are restricted to the infant and juvenile forms of the disease.
  - There is one large case series relating to the adult-type disease, but it is in the Orthopedics literature and only mentions hearing loss in passing.
  - There are two similar case reports in the international Otolaryngology literature relating to adult-type osteopetrosis and its otologic consequences.

- Current research is focusing on the sclerostin molecule as a possible target for treatment, but the prognosis for the infantile and juvenile types remains poor.

Case Report

- Our patient first presented in 1992 at the age of 37 with complaints of bilateral tinnitus. Of note he had a history of unilateral facial nerve paralysis which was treated with surgical decompression in 1980. Physical examination at that time was pertinent for severe rhinitis, bilateral serous otitis media, and an equivocal Rinne exam with a 512Hz tuning fork. Audiometry was reported as a mild (25-30dB) conductive hearing loss bilaterally with flat tympanograms. He was treated conservatively at that time with nasal steroids.

- The patient returned in 1995 with complaints of increased tinnitus bilaterally. The Weber exam lateralized to the right and bone conduction was greater than air conduction on that side. Audiometry confirmed our tuning fork studies with progression of his conductive hearing loss, worse on the right than the left. A temporal bone CT was ordered which was found to be consistent with osteopetrosis tarda. The patient was fitted for hearing aids and referred for genetic counseling.

- By 2007 the patient had progressed to a moderate to severe mixed-type hearing loss. He continues to be managed with amplification devices. We continue to follow the progression of his sclerotic disease with serial audiograms and CT scans. His most recent CT images are seen to the left.

Discussion

- Osteopetrosis is a very rare congenital cause of hearing loss in both the infantile and adult population.

- We attribute the conductive hearing loss in our patient to sclerotic fixation of the ossicles as well as chronic serous otitis media due to bony obstruction of the eustachian canal.

- The sensorineural component is attributed to bony stenosis of the internal auditory meatus with resultant CN VIII compression.

- There is a relative paucity of information in the otolaryngology literature about the adult-type presentation of osteopetrosis.

- While most practitioners may never come across a patient with this rare disorder (particularly of the adult type) it is nonetheless important for us to keep informed of the manifestations and presentation of this unusual cause of mixed-type hearing loss.

Images

Figure 1.a: axial cut of a non-contrast Temporal Bone CT scan through the level of the external auditory canals demonstrating near total sclerosis of the mastoid air cells and maxillary sinuses.

Figure 1.b: coronal cut from the same non-contrast CT scan moving posteriorly from the external auditory canals through the sclerosed mastoid air cells.

Figure 2.a and 2.b are audiograms from the patient at presentation in 1992 (a) and more recently in 2007 (b) showing progression from a mild conductive loss to a moderate-severe mixed loss.