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Brachio-oto-renal syndrome: CT imaging and intra-operative diagnostic findings

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Branchio-oto-renal syndrome (BOR) is an autosomal dominant mutation of the EYA1 and the more recently discovered the SIX1 gene.¹ The phenotype and syndrome were comprehensively described by Melnick in 1975 to include hearing loss, auricular malformations, branchial arch remnants, and renal anomalies.² The diagnosis of BOR is made using major and minor criteria as defined by Chang et al 2004.³ However, 60% of patients who meet phenotypic criteria do not have an identifiable mutation in the EYA1 gene, leading to recent interest in the EYA-SIX regulatory system.¹

The most common manifestations include hearing loss (98.5%), preauricular pits (83.6%), branchial anomalies (68.5%), renal anomalies (38.2%), and external ear abnormalities (31.5%). In terms of the imaging characteristics, the most sensitive modality remains CT of the temporal bones. The most commonly reported anomalies on temporal bone imaging include but are not limited to 1) hypoplastic apical turn of the cochlea, 2) facial nerve deviated to the medial side of the cochlea, 3) funnel-shaped internal auditory canal, and 4) patulous eustachian tube.⁴

The spectrum of hearing loss in BOR is variable but most commonly presents with mixed hearing loss (50%), pure sensorineural hearing loss (25%) and pure conductive hearing loss (25%)⁵. The conductive component of the hearing loss is most often the result of ossicular chain abnormalities.

A 42 year old male previously diagnosed with BOR using clinical criteria presented with a conductive hearing loss. His physical exam demonstrated small external auditory canals with a normal tympanic membrane. His audiogram demonstrated a mild left sensorineural hearing loss and a maximal conductive hearing loss in the right ear.

Imaging with CT revealed several findings consistent with BOR: Bilateral enlarged air-filled eustachian tubes extending from the middle ear to the nasopharynx, a widened and flared internal acoustic meatus with the nervus intermedius extending into a funnel shaped

labyrinthine segment of the temporal bone and hypo-plastic horizontal canal, hypo-plastic vestibular system/epitympanum, and lateral position of the facial nerve. The incus and malleus were malformed and fixed in the attic (Fig. 1).

Despite the findings on CT, the patient elected to pursue a middle ear exploration prior to pursuing other rehabilitative options. At surgery, middle ear exploration revealed a very small oval window niche with no clear oval window, or stapes footplate as shown on this view with a 30° endoscope (Fig. 2). The round window niche was visible. A dehiscence of the facial nerve was visible at the horizontal segment. Because no mobile footplate was found there was no attempt at ossiculoplasty. The patient recovered from surgery with no change in his hearing and later went on to a Baha which he found beneficial.

The extreme ossicular abnormalities in this patient with BOR made his maximal conductive hearing loss not amenable to ossiculoplasty. We ultimately failed in our attempt to restore his conductive hearing loss due to agenesis of the oval window and lack of a mobile footplate. The intraoperative endoscopy revealed the lack of suitable anatomy for an ossicular replacement prosthesis. The findings in our patient highlight the diagnostic findings in BOR on CT scan of the temporal bone and correlate well with his clinical findings. Although BOR patients may not be a homogeneous group in terms of their middle ear anatomy, this patient suggests ossiculoplasty may not be a viable option in this population.

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Figure 1.

Computed tomography (CT) images. Panel A: Axial view of the head demonstrating enlarged eustachian tubes (arrow). Panel B: Axial view of right internal auditory (IAC) canal demonstrating hypoplastic horizontal semicircular canal(H), and funnel shaped labyrinthine segment of the facial nerve (arrow). Panel C: Coronal view of temporal bone demonstrating lateral facial nerve (arrow), and hypo-plastic vestibular organs (v). Panel D: Axial view of right temporal bone showing the incus and malleus are malformed and fixed in the attic (arrow) and eustachian tube (ET).

Figure 2.

Right middle ear as viewed through a 30° endoscope during surgery. There was no clear stapes footplate, but a narrow oval window (OW) niche. The round window (RW) niche was visible. The OW and RW are labeled to the right above the structures. The horizontal segment of a dehiscent facial nerve (F) is clearly visible. The remnants of the incus (I) and malleus (M) are marked.