


Multimodality depiction of findings in branchio-oto-renal syndrome: two case reports

Acta Radiologica Open
8(7) 1–3
© The Foundation Acta
Radiologica 2019
Article reuse guidelines:
sagepub.com/journals-permissions
DOI: 10.1177/2058460119861606
journals.sagepub.com/home/arr


Lelia E Williams, Mark T Sohner, Robert C Sibley III and
Cory M Pfeifer 

Abstract

Branchio-oto-renal syndrome is a rare genetic disorder that affects multiple organ systems. Temporal bone abnormalities include the unwound appearance of the cochlea which is common in this syndrome. This appearance can prompt renal imaging and evaluation. Presented here are two cases of branchio-oto-renal syndrome with dysplastic cochleae. A branchial cleft sinus and renal dysplasia were also present in one of the cases.

Keywords

Branchio-oto-renal syndrome, cochlear dysplasia, magnetic resonance imaging

Received 18 March 2019; accepted 14 June 2019

Introduction

Branchio-oto-renal syndrome (BORS) is characterized by the abnormal development of the second branchial arch, ears, and kidneys. Patients commonly present with hearing loss which can be conductive, sensorineural, or mixed (1). There can also be outer, middle, and/or inner ear malformations. Other symptoms include branchial cleft cysts and fistulae in the neck as well as abnormalities of kidney structure and function. These range from mild to severe and may affect one or both kidneys.

Here we present two cases of BORS as a means to facilitate diagnosis and prompt appropriate screening in similar patients.

Case report 1

A 13-month-old girl presented with bilateral sensorineural hearing loss, bilateral eustachian tube dysfunction, a patent ductus arteriosus, and chronic lung disease. Her family history was significant for multiple relatives exhibiting preauricular pits. On physical exam, she displayed preauricular pits, a submandibular pit, dolichocephaly, micrognathia, and low set ears.

Magnetic resonance imaging (MRI) of the internal auditory canals demonstrated an unwound appearance of both cochleae (Fig. 1a) with abnormal cystic coalescence of the middle and apical turns bilaterally. Sagittal

reformats through both internal auditory canals showed small caliber cochlear nerves (Fig. 1b).

A palpable pit on the right neck was suspected to represent a branchial sinus. A subsequent fistulogram performed at the age of two years (Fig. 1c) revealed contrast tracking along the anterior medial margin of the right sternocleidomastoid muscle extending medially at the level of the angle of the mandible where it emptied into the lateral aspect of the oropharynx near the tonsillar fossa consistent with a type II branchial anomaly.

The left kidney was not seen on ultrasound; however, there was minimal renally active tissue present in the left renal fossa appreciable via renal scintigraphy (Fig. 1d).

Case report 2

A three-year-old girl presented to an otolaryngologist with hearing loss. Her surgical history was significant

Department of Radiology, University of Texas Southwestern Medical Center, Dallas, TX, USA

Corresponding author:

Cory M Pfeifer, Department of Radiology, University of Texas Southwestern Medical Center, 5323 Harry Hines Boulevard, Dallas, TX 75390, USA.

Email: Cory.Pfeifer@utsouthwestern.edu

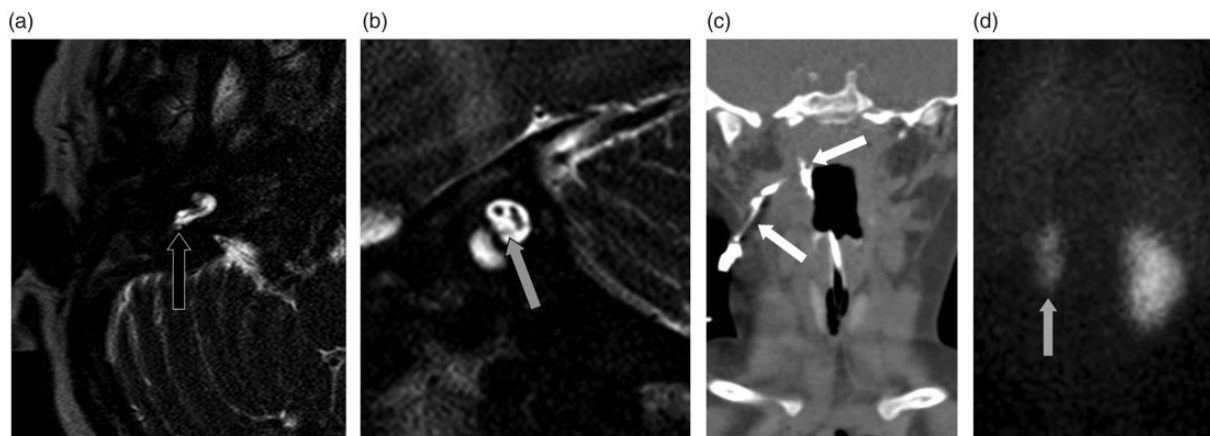


Fig. 1. Case 1. (a) Axial high resolution heavily T2-weighted balanced fast field echo image shows the “unwound” appearance of the cochlea (black arrow with white outline) with an elongated basal turn. (b) A sagittal reformatted image demonstrates a hypoplastic cochlear nerve (gray arrow with black outline). (c) Coronal reformatted image from a CT scan of the neck was obtained following injection of 5 mL Isovue-300 injected into a small opening on the right anterior neck. Contrast tracks through the branchial sinus as denoted by the white arrows. 1.00 mCi mercaptuacetyltriglycine (MAG3) technetium-99m was injected and continuous scintigraphy was performed. (d) The 2-min frame is shown. The left kidney is hypoplastic compared to the right. The differential renal function was computed as 85% on the right and 15% on the left based on the 2-min image.

for prior bilateral branchial cleft cyst excision. She was born to a methamphetamine-addicted mother, and her past medical history was significant for neglect. She and her sibling were removed from the custody of their biologic parents and placed in foster care. On physical exam, her bilateral external auditory canals were impacted with cerumen. Her pinnae were normal. A mild left facial palsy was present and strabismus was observed.

Computed tomography (CT) of the temporal bones revealed multiple abnormalities (Fig. 2). On the right, there were multiple fixation points between the malleus and the incus. A normal stapes was not observed. The cochlea exhibited an unwound configuration with a lengthened basal turn. The left temporal bone likewise showed multiple fixation points between the malleus and the incus as well as an absent stapes. The unwound appearance of the left cochlea was similar to the right. Both facial nerves exhibited abnormal lateral courses.

A renal ultrasound in this patient was unremarkable with no abnormality demonstrated.

Discussion

BORS is an autosomal dominant disorder affecting approximately 1 in 40,000 people and is caused by mutations in three genes: EYA1; SIX1; and/or SIX5 (2). Though most cases are inherited, the disease may arise from a random mutation. Within the affected population, 40% of the disease is caused by a mutation in the EYA1 gene (3). EYA1 encodes a transcriptional coactivator that is important in the fetal development of the branchial arches, ears, and kidneys. However,

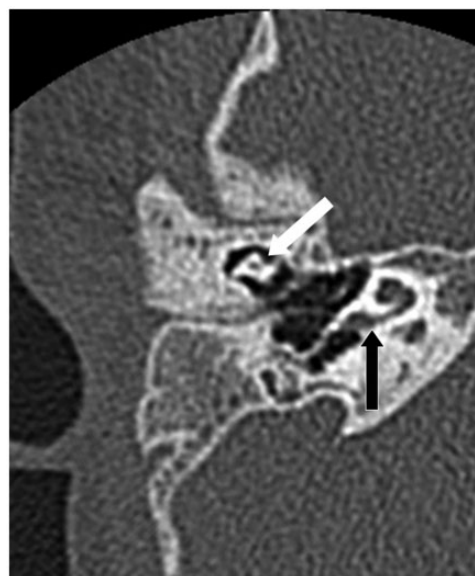


Fig. 2. Case 2. Axial CT of the right temporal bone in a three-year-old girl shows dysmorphic ossicles with fusion of the incus and malleus (white arrow). The short process of the incus is fused to the lateral recess of the tympanic cavity. An unwound cochlea is depicted by the black arrow.

phenotypic expression of BORS is variable in presentation and severity, even among individuals in the same family.

There are many genetic causes for hereditary hearing loss, some of which are associated with characterized syndromes. Though a rare cause of hearing loss, a diagnosis of BORS has important implications not only for the individual, but for the health of the family.

Multiple middle ear anomalies have been reported in BORS. The unwound appearance of the cochlea in these patients holds a specific association with BORS (4) and can prompt neuroradiologists to suggest specific testing. This finding in an undiagnosed patient should also prompt renal imaging because of the strong association with kidney malformation and potentially life-threatening renal dysfunction in this condition.

Branchial cleft cysts, fistulas, and outer ear malformations may necessitate surgical correction. Surveillance for otologic and renal anomalies is often performed. Typical surveillance protocols include annual otoscopy, semi-annual exam for hearing impairment, and annual audiometry to assess stability of hearing loss. Also typical are recurring exams by nephrologists and/or urologists.

The patients detailed here presented with complex constellations of signs and symptoms that suggested a syndromic etiology; however, the severity and presentation of BORS can vary wildly. This can complicate the diagnosis in milder cases. Since most patients initially present with symptoms of hearing loss, the unwound appearance of the cochlea on imaging may provide an important clue to making the diagnosis of BORS.

In conclusion, BORS is a rare disease that produces abnormalities in multiple organ systems. Neuroradiologists may be the first to suggest this disorder and prompt additional testing as needed.

Declaration of conflicting interests

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The author(s) received no financial support for the research, authorship, and/or publication of this article.

ORCID iD

Cory M Pfeifer  <https://orcid.org/0000-0002-2220-9230>

References

1. Stinckens C, Standaert L, Casselman JW, et al. The presence of a widened vestibular aqueduct and progressive sensorineural hearing loss in the branchio-oto-renal syndrome. A family study. *Int J Pediatr Otorhinolaryngol* 2001;59:163–172.
2. Morisada N, Nozu K, Lijima K. Branchio-oto-renal syndrome: comprehensive review based on nationwide surveillance in Japan. *Pediatr Int* 2014;56:309–314.
3. Chang EH, Menezes M, Meyer NC, et al. Branchio-oto-renal syndrome: the mutation spectrum in EYA1 and its phenotypic consequences. *Hum Mutat* 2004;23:582–589.
4. Hsu A, Desai N, Paldino MJ. The unwound cochlea: a specific imaging marker of branchio-oto-renal syndrome. *AJNR Am J Neuroradiol* 2018;39:2345–2349.