

CASE REPORT

BRANCHIO-OTO-DYSPLASIA SYNDROME: A RARE PRESENTATION OF BRANCHIAL CLEFT ANOMALIES

Kulkarni Manik Rao¹

HOW TO CITE THIS ARTICLE:

Kulkarni Manik Rao. "Branchio-Oto-Dysplasia Syndrome: A Rare Presentation of Branchial Cleft Anomalies". Journal of Evidence based Medicine and Healthcare; Volume 1, Issue 13, December 01, 2014; Page: 1671-1674.

ABSTRACT: We report a rare case of bilateral branchial fistulae, bilateral preauricular sinuses with bilateral conductive hearing loss, called "the branchio-oto-dysplasia syndrome" (BO syndrome). It is considered as a variant of branchio-oto-renal syndrome (BOR syndrome), in which renal anomalies is an added feature. The incidence of hearing loss and the renal anomalies as per literature is variable. It has to be kept in mind that, a case with branchial fistula can be associated with various other anomalies like preauricular pits, hearing loss, and rarely renal anomalies also.

KEYWORDS: Branchial, oto, dysplasia, anomalies.

INTRODUCTION: The branchio-oto-dysplasia syndrome (BO syndrome), is an uncommon autosomal dominant disorder, classically presents with branchial fistula, preauricular sinuses and hearing loss. It may be associated in few cases with renal abnormalities, when it is considered as branchio-oto-renal syndrome (BOR syndrome).

On the basis of literature and clinical observation, the presentation of cases without renal anomalies should not be considered as a different entity, but a variant of BOR syndrome. Here, we discuss a variant with bilateral branchial fistula, bilateral preauricular sinuses and bilateral conductive hearing loss, without renal anomalies, said to be the branchio-oto-dysplasia syndrome.

CASE REPORT: A 6 year old girl presented to ENT outpatient department, with two small openings on either side of the neck, at the lower one third of the anterior border of sternocleidomastoid muscle (Fig 1& 2), with serous fluid oozing out, since birth. Patient also had bilateral preauricular pits with bilateral hearing loss. Otologic examination revealed normal appearing and mobile tympanic membrane on both sides. Fistulogram of bilateral surgical fistulae revealed a blind left fistulous tract and the right tract opening in the pyriform sinus internally. Audiogram revealed bilateral conductive hearing loss. Routine investigations were normal. Specific investigations revealed no renal abnormalities. No other family member had similar findings.

The cervical fistula and preauricular sinuses on both sides were removed surgically under general anaesthesia. The left cervical fistula was about 3.5 cm long blind tract, which was removed by a step ladder incision. The right cervical fistula passed between internal and external carotid arteries and then was traced till the pyriform fossa internally into the right pyriform fossa. The whole tract was excised. Both preauricular sinuses were removed. All the fistulous tracts and the sinuses were sent for histopathological examination separately.

CASE REPORT

Histopathological examination report showed stratified squamous epithelium with underlying lymphoid follicles below the epithelium, confirming the diagnosis of branchial tract fistula and preauricular sinuses (Fig 3). Bilateral conductive hearing loss was corrected by ossiculoplasty subsequently in the second settings. Long term follow up of the patient show correction of all the defects.

DISCUSSION: In a two week embryo, branchial apparatus consists of five branchial arches, five clefts and five pouches. Failure of branchial pouches to obliterate in uterus results in cysts or sinus tracts.

The second arch is the most commonly malformed. A branchial fistula is caused by persistence of a sinus external opening. It consists of a skin lined tract, opening anywhere in the oropharynx or hypopharynx, commonly being on the anterior aspect of the tonsillar fossa or pyriform fossa, and the external opening at the anterior border of sternocleidomastoid muscle, at the junction of middle and lower one third.

The first report of combination of preauricular pits, branchial fistulae and hearing impairment was by Heusinger in 1864. Fara¹ et al described a family with hereditary deafness, branchial cleft anomalies and incidentally found one member of the family to have unilateral renal agencies. It was as early as 1927; Precechtel² reported several members of a family over four generations manifested branchial anomalies and hearing loss. It was Melnick³ in 1976, first described BOR syndrome as an AD disorder consists of branchial cleft, renal, and otologic anomalies with associated hearing loss, earlier to which BO syndrome and ear pit deafness syndrome were considered to be distinct entities.

The estimated prevalence of BOR syndrome is one in 40,000 live births. The typical phenotype expression includes hearing loss, preauricular pits, and branchial cleft anomalies.⁴ Even though the gene responsible is having high penetrance, the expression is variable. All the features of the syndrome are not expressed in all the carriers of the gene.⁵

The otologic finding in BOR syndrome may include external, middle and inner ear anomalies. 75% of cases with BOR syndrome have some type of hearing loss, with 50% being mixed hearing loss, 20% sensorineural hearing loss and 30% conductive hearing loss. The preauricular pits are usually shallow and end in blind depression just anterosuperior to the upper attachment of helix of the ear. Most of the renal anomalies are minor and asymptomatic but an estimated 6% incidence of severe renal dysplasia is reported.⁶

Isolated branchial fistulae are found in 6% in males, 40% in females. Sinuses are often noted at birth. In 60% of cases it is found left side, 40% on the right side and around 6% cases bilaterally.

CONCLUSION: We present a rare form of presentation of bronchial cleft anomalies said to be branchio-oto-dysplasia syndrome. Though this condition is familial, it can be presented as an isolated case as in our study. As an ENT surgeon, it has to be kept in mind that, a case with branchial fistula can be associated with various other anomalies like preauricular pits, hearing loss, and rarely renal anomalies also. These associated anomalies have to be looked for, properly investigated and managed.

CASE REPORT

BIBLIOGRAPHY:

1. Fara M, Chlupackova V, Hrivnakova J., "Dismorphia oto-facio-cervicalis familiaris", Acta chirip last. 1967; 9: 255-268.
2. Precechtel A., "Pedigree anomalies in the first and second branchial cleft". Acta otolaryngol (stockh). 1927; 11: 23-30.
3. Melnick M, Bixler D, Nance W. E, et al., "Familial branchio Oto renal dysplasia: a new addition to the branchial arch syndrome", clin genet. 1967; 9: 25-34.
4. Millman Brad, Gibson Williams S, Foster wayne P, "Branchio-oto-renal syndrome", Archives of otolaryngology and head and neck surgery. aug-1955;121 (8):922-925.
5. Rana I, Dhawan R, Gudwani S. et al., "Non inherited manifestation of bilateral branchial fistulae, bilateral preauricular sinuses and bilateral hearing loss: a variant of branchio-oto-renal syndrome", Indian J otolaryngol head neck surgery, Jan-march 2005;57(1).



Fig. 1: Bilateral Branchial Fistulas



Fig. 2: Preauricular Pit Along With Branchial Fistula

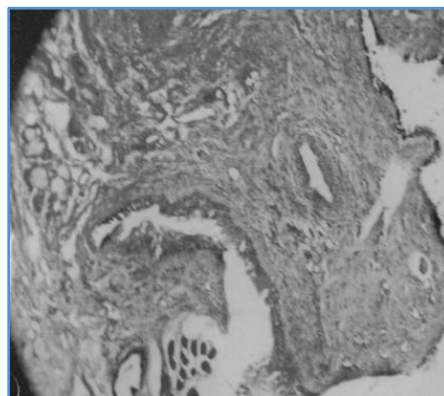


Fig. 3: Histopathological Examination Finding Of Excised Fistulous Tract

CASE REPORT

AUTHORS:

1. Kulkarni Manik Rao

PARTICULARS OF CONTRIBUTORS:

1. Associate Professor, Department of ENT, VIMS, Bellary, Karnataka.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Kulkarni Manik Rao,
57, Yannareddy Compound,
1st Cross, Infantry Road,
Bellary, Karnataka-583104.
E-mail: drmanvrk@rediffmail.com

Date of Submission: 07/11/2014.
Date of Peer Review: 08/11/2014.
Date of Acceptance: 11/11/2014.
Date of Publishing: 28/11/2014.