CASE REPORT

Coexistence of bilateral first and second branchial arch anomalies

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SUMMARY

Branchial arch anomalies are one of the most common congenital anomalies that are usually unilateral and bilateral presentation is rare. The simultaneous presence of bilateral second branchial arch anomalies along with bilateral first arch anomalies is extremely rare, with only three such cases reported in the literature. We present two non-syndromic cases of coexisting bilateral first and second arch anomalies. Developmental anomalies of the branchial apparatus account for 17% of all paediatric cervical masses and are the most common type of congenital cervical mass. They usually present in the paediatric age group. About 96–97% of these anomalies are unilateral. Bilateral presentation is seen in 2–3% having a strong familial association. Congenital syndromes also have been associated with first and second branchial arch anomalies. Thorough clinical examination and investigations should be done to rule out these syndromes.

BACKGROUND

A branchial anomaly is a congenital developmental defect that occurs due to the failure of obliteration of the primitive branchial apparatus.¹ Almost 90% of these branchial anomalies are attributed to the second arch anomalies.^{2–3} Bilateral presentation of branchial anomalies is rare, seen in about 2–3% of all cases. Mostly, such cases have a familial component.⁴ Preauricular sinus is a common first arch anomaly seen bilaterally in 25–50% of the presentations. However, the simultaneous presence of bilateral first and second arch anomalies is extremely rare. We searched the literature (PubMed, MedLine and Google Scholar) on this rare presentation and found only three such cases.^{5–7}

Figure 1 Left side view of case 1 showing pin-point opening in the preauricular area and cyst in the neck on left side.

Figure 2 Right side view of case 1 showing pin-point opening in the preauricular area and neck on right side.

We present two new cases of bilateral second branchial arch anomaly coexisting with bilateral first arch anomaly.

CASE PRESENTATION**Case 1**

A 17-year-old male patient presented with bilateral pinpoint openings in lower one-third of neck with intermittent discharge from right side since birth. Further history was non-contributory. On examination (figures 1 and 2), 0.1×0.1 cm size openings were present at the junction of upper two-third and lower one-third of anterior border of sternocleidomastoid muscle (SCM), bilaterally. Examination also revealed 0.1×0.1 cm asymptomatic openings in front of both ear. Further, clinical examination was normal. A probable diagnosis of first and second arches was kept, and the patient was subjected to further investigations.

Case 2

An 8-year-old female patient presented with discharge from bilateral pin point openings in the lower one-third of the neck since birth. She also had discharge from bilateral pin point openings in front of the ear since birth. Other history was non-

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contributory. On examination (figures 3–5), there were 0.1×0.1 cm openings present at the junction of upper two-third and lower one-third of anterior border of SCM bilaterally with purulent discharge on palpation. Patients also had discharge and excoriation around 0.1×0.1 cm openings in front of both ears.

INVESTIGATIONS

Case 1

The ultrasonography (USG) of the neck found a small sinus of 1 cm thickness on the right and a well-encapsulated 2×2 cm cyst on the left anterior triangle of neck along the SCM anterior to the common carotid artery (CCA) and internal jugular vein. Ultrasound abdomen and haematological investigations were normal.

Case 2

USG of the neck was not performed due to infection and USG abdomen did not find any abnormality.

DIFFERENTIAL DIAGNOSIS

We kept the first possibility of coexisting bilateral first and second brachial arch anomalies. The diagnosis of this anomaly is usually based on characteristically clinical features described in

these case reports. However, radiological investigations can delineate the disease better.

TREATMENT

Case 1

Excision of bilateral preauricular sinuses, right branchial sinus and left branchial cyst was done under general anaesthesia.

Case 2

The patient was put on conservative management due to infected sinus, and advised regarding regular follow-up.

OUTCOME AND FOLLOW-UP

The histopathological examination found sinus tracts lined by stratified squamous epithelium with sebaceous and seromucinous glands. The excised preauricular sinus also had fibrocartilaginous and adipose tissue. Postoperative period and single

Figure 3 Clinical photograph showing pin point opening with purulent discharge on both side of neck.

Figure 4 Left side view of case 2 showing pin point opening in the preauricular area.

Figure 5 Right side view of case 2 showing pinpoint opening in the preauricular area with adjacent ulcer.

follow-up after 3 months was uneventful in case 1. Infection was controlled in case 2, and has been planned for surgery.

DISCUSSION

Development and differentiation of the primitive branchial apparatus occurs in the second and seventh week of gestation. Initially, five ridges appear on the ventrolateral surface of the embryonic head. These ridges form the branchial arches. Each arch consists of a core of mesenchyme, and is covered externally with ectoderm, representing the branchial cleft, and internally with endoderm, representing the branchial pouch. These clefts and pouches approach each other to form a closing membrane. Invasion of the surrounding mesenchyme gradually obliterates the cleft and pouch in humans.

With further development, the second arch first increases in size and proliferates caudally to meet the enlarging ridge of the fifth arch. This results in the formation of an enclosed ectodermally lined cavity, the cervical sinus of His. Later, the sinus obliterates by the apposition and fusion of its walls. Failure of this obliteration results in branchial anomalies.

Branchial arch anomalies can arise from first, second, third or fourth arch. First arch constitutes 8–10% of branchial anomalies.⁸ WP Work⁹ described two types of first branchial anomalies. In type I anomalies, there is duplication of the external auditory canal, which open in the preauricular area. In type II anomalies, the tract is in close association with the parotid gland and the facial nerve. They open posterior or inferior to the angle of mandible.

The second arch contributes 95% of the total branchial arch anomalies.¹⁰ These anomalies are found along the anterior border of the SCM. Its tract lies between the internal and external carotid arteries, lateral to the CCA, the glossopharyngeal nerve and the hypoglossal nerve. The sinus opens into the region of tonsillar fossa and normally ends close to the middle constrictor muscle.¹¹

The third arch anomalies have an incidence of 2–8%.^{11 12} They also appear at the anterior border of SCM. Its tract ascends lateral to the CCA, superior to the hypoglossal nerve and inferior to the glossopharyngeal nerve. It terminates by piercing the lateral thyroid membrane and opens into the pyriform sinus.¹¹

The fourth branchial arch anomalies, which are extremely rare, account for about 1–2%.^{11 12} They travel along the ascending recurrent laryngeal nerve below the arteries of the fourth arch (aortic arch, subclavian artery), and open in the upper oesophagus.¹³

Developmental anomalies of the branchial apparatus account for 17% of all paediatric cervical masses and are the most common type of congenital cervical mass.¹⁴ They are usually present in the paediatric age group. About 96–97% of these anomalies are unilateral. Bilateral presentation is seen in 2–3%, with a strong familial association.⁴ Congenital syndromes that have been associated with first and second branchial arch anomalies include Oculoauricularvertebral dysplasia (Goldenhar syndrome), Branchio-oto-renal syndrome, Treacher-collins syndrome, Pierre-Robinson sequence and Auriculocondylar syndrome.¹⁵ A thorough clinical examination should be done and if suspected, proper investigations must be carried out to rule out these syndromes.

We did not find any family history or associated syndrome in these two cases. The diagnosis of first and second branchial anomaly is usually based on characteristic clinical features as investigations are contentious.¹⁶ Preoperative contrast studies delineate the sinus tract well and help in surgical approach.¹⁷ Sometime, we perform contrast x-rays (sinugram), and ultrasound are done in cases with cyst formation. We do not perform expensive radiological investigations like CT/MRI scan in first and second branchial anomaly; however, they are helpful in branchial cyst, third/fourth branchial arch anomaly^{18 19} and associated neck abscess and syndromes. Furthermore, in our opinion, radiation exposure in CT scan should also be taken into account over its advantage in first and second branchial sinus.

We treat the infective sinus by systemic penicillin preferably amoxycyclavunate. Chronic discharging sinus needs bacterial culture and exclusion of tuberculosis especially in patient belonging to tubercular endemic areas like India. However, a thorough history and routine investigations like complete blood count usually give hint to tubercular pathology. We did not suspect tuberculosis in second case as it was the first episode of infection and history ruled out any tubercular exposure.

Open or endoscopic surgical excision is the standard treatment for these anomalies.¹⁶ Other modalities include endoscopic electrocoagulation²⁰ and chemical cautery²¹ but all these surgical modalities carry few complications especially in paediatric age group.^{16 21 22} We prefer single stage open surgery in

these bilateral, multiple anomalies due to cost effectiveness until and unless patient/parents disagree. Peroperatively, we delineate the tract with coloured dye to assess its course. Preauricular sinuses are excised similar to the technique described by Bae *et al.*²³ A complete second branchial sinus is excised through step-ladder incisions with favourable cosmetic outcome. We have not encountered any complication or recurrence in open surgical method.

Learning points

- ▶ Simultaneous presentation of first and second arch anomalies is extremely rare, and only three cases with similar findings have been reported.
- ▶ There can be bilaterality, simultaneous another branchial arch anomaly or congenital syndromes associated with branchial arch anomalies.
- ▶ Patients of branchial arch anomalies should be examined and investigated in detail to rule out associated disease.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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