Case Report

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Anomalies of the branchial arch apparatus in children: case series and review of literature

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ABSTRACT

Branchial apparatus develop between the 3rd and 7th weeks of embryonic life. There are five mesodermal arches separated by invaginations of ectoderm and endoderm. Branchial anomalies are result of aberrant embryonic development and are rarely seen in clinical practice. The objective of this article is to present few cases of branchial arch anomalies and to discuss the clinical presentation, diagnosis and surgical approach of such lesions in pediatric age group.

Keywords: Branchial arch anomalies, Pediatric age, Branchial apparatus, Branchial fistula, Branchial cyst

INTRODUCTION

Branchial arch anomalies (BA) are the second most common head and neck lesions seen in pediatric age group. Six pairs of branchial arches appear in the fourth week of embryonic life and they have a vital role in the development of face, neck and pharynx.^{2, 3} The fifth arch disappears and the sixth arch is rudimentary.3 Each branchial arch is made up of a core of mesenchymal tissue, covered externally by ectoderm and internally by endoderm. During development the second (II) arch grows caudally and combines with third (III) and fourth (IV) pharyngeal arches to form ectodermal lined cavity called cervical sinus of His. Generally, this cervical sinus rapidly involutes during embryogenesis, absence of this process leads to branchial arch anomalies. Depending on the anatomic location, branchial anomalies are classified into first, second, third, and fourth arch anomalies. 4 These anomalies present as cysts, sinus tracts, fistulae and occasionally also as cartilaginous remnants. Around 20% of cervical masses in children are branchial anomalies.¹

In this article we are presenting a case series of branchial arch anomalies encountered in our department of

pediatric otorhinolaryngology. Detailed information on the characteristic clinical features, radiological and pathological diagnosis, with its management is discussed. An attempt is made to throw light on the review of literature of this vast topic.

CASE REPORT

Case 1: Collaural fistula (First branchial cleft anomaly)

A 2.5 year old girl, presented to us with the chief complaints of a discharging pit in the left side of upper neck since two years. On clinical examination, (Figure 1a) an opening was identified in the left submandibular triangle, 2 cm from the angle of mandible with active pus discharge. Differential diagnosis of first branchial arch anomaly and orodentocutaneous fistula made. Pus sent for culture and sensitivity and oral antibiotics started. AFB staining of the same found to be negative. No osteomyelitic changes seen on OPG. The child was taken up for surgical excision after routine investigations and anesthetic clearance. Methylene blue dye injected into the fistulous opening. An elliptical incision made at the site of the opening and tract traced into the parotid tissue

lying superficial to facial nerve and extending into the cartilaginous part of the external auditory canal (Figure 1b). Tract with a piece of cartilage of the external auditory canal removed in toto. The wound was closed in

two layers. The excised specimen sent for histopathological examination and was reported as well epithelised sinus tract lined by keratinised stratified squamous epithelium.

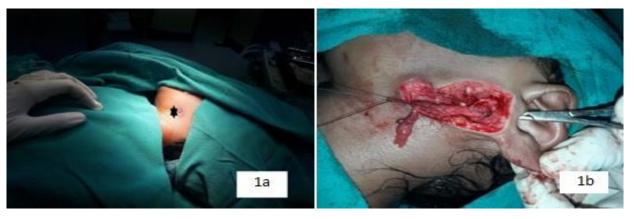


Figure 1: First branchial cleft anomaly. (A): A discharging pit (*) in upper left side of neck; (B): tract lying in parotid tissue and extending into left external auditory canal.

Case 2: Second arch branchial fistula

A 5 year old boy with congenital intermittent discharging sinus in the left lower neck was examined and spot diagnosis of II arch branchial fistula was made in view of classical site and clinical presentation. Parents counselled regarding surgery. The child was taken up for surgical excision after routine investigatons and anesthetic clearance. Methylene blue dye injected into the fistulous opening and intraoral spurge of dye seen in left tonsillar region. Elliptical incision made around the sinus opening and tract traced. Step ladder incision made at level of the hyoid in upper neck crease and the dissected tract burrowed through it. Tract was seen passing between the carotid bifurcations and closed to hypoglossal nerve, and dissected out carefully till the tonsillar area and completely excised. Wound was closed in layers. It was reported as epithelized tract lined by keratinised stratified squamous epithelium (Figure 2).

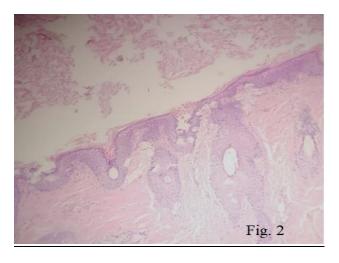


Figure 2: Photomicrograph of a sinus.

Case 3: Type I second branchial cyst and second arch branchial fistula

A 5 year old girl presented to us with chief complaints of painless swelling on right side of the neck and intermittent discharging opening on the left side of the neck from last one year (Figure 3a). On examination, swelling of size 2 cm x 1.5 cm was seen on right side of neck, anterior to the lower third of sternocleidomastoid (SCM) muscle. On palpation, swelling was non tender, cystic and becomes more prominent with contraction of the ipsilateral SCM muscle. On the left side of the neck, a small dry opening was present along the anterior border of the SCM muscle, at the junction of upper 2/3 rd and lower 1/3rd of muscle. FNAC from right neck swelling yielded pale yellow fluid and reported as benign development cyst.

After routine investigation and pre anesthetic clearance, child underwent surgical excision of right sided neck cyst and left neck fistula under general anesthesia. Cyst was found in the subplatysmal plane, lying deep to the SCM muscle and extending as fistulous tract in between internal and external carotid artery (Figure 3b). The cyst with fistulous tract was traced upto the tonsillar area, completely excised in toto and sent for histopathological examination.

On left side, we injected methylene blue dye to identify the fistula tract and dye did not appear in the tonsillar region suggestive of obliterated tract. An elliptical incision was made over the skin opening and the tract was dissected till tonsillar area (Figure 3c) and excised, upper end of the tract was ligated to prevent further recurrence. The wound was closed in layers. A cyst lined by ciliated columnar epithelium on right side and a tract with squamous epithelium lining was reported by pathologist (Figure 4).



Figure 3 (A): An ill defined swelling of size 2 cm×1.5 cm on lateral aspect of right neck and an opening along the anterior border of SCM muscle, on left side; (B): cyst with tract extending up to right tonsillar fossa; (C): tract passing through carotid bifurcation and reaching till tonsillar region; (D): gross specimen of cyst and ligated left sided sinus tract.

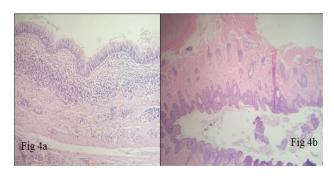


Figure 4: Photomicrograph of a cyst and sinus. (A): Cyst wall lined by pseudostratified ciliated columnar epithelium, subepithelium shows dense mixed inflammation (H & E stain, 100 X);
(B): photomicrograph of a well epithelialized sinus tract. Lining epithelium shows stratified squamous epithelium (H & E stain, 100 X).

Case 4: Type II second branchial cyst

Six year old girl reported to us with gradually increasing left sided neck fullness since last 6 months. Examination revealed diffuse ill-defined mass in upper left side of the neck. On palpation a soft to cystic non-tender swelling of

size 4 cm×3 cm was found deep to upper 1/3 rd of left SCM muscle. Rest of neck examination was normal. On aspiration, straw coloured fluid came out and cytology was consistent with branchial cyst. Contrast MR imaging of neck (Figure 5c) revealed a well defined cystic lesion in left submandibular region lateral to carotid space, measuring 29×28×52 mm. The non enhancing lesion was situated postero-lateral to carotid bifurcation and anteromedial to sternocleidomastoid muscle. Thus provisional diagnosis of type II second arch branchial cleft cyst was made and surgery was planned. Skin incision was given in upper neck crease, over the cyst. Superior and inferior subplatysmal flaps were raised, and SCM muscle retracted to visualize the mass. The cystic mass (Figure 5a) seen lying in the carotid sheath in between the carotid and internal jugular vein. The cyst (Figure 5b) carefully separated from the surrounding structures and from the adherent vagus nerve, without damaging the wall and wound closed in layers. Postoperative period was uneventful. Diagnosis of branchial cleft cyst confirmed on histopathology report (Figure 6).

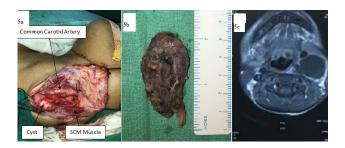


Figure 5: Type II branchial cyst. (A): cyst lying in carotid sheath, lateral to carotid artery; (B): gross specimen of cyst with few adhered lymph nodes; (C): Contrast MR imaging of neck showing a well defined cystic lesion, measuring 29×28×52 mm, situated just below the angle of mandible and posterolateral to the carotid bifurcation and anteromedial to sternocleidomastoid muscle.

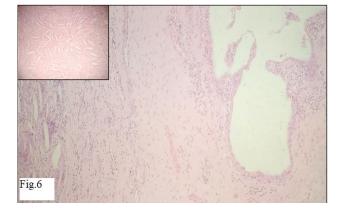


Figure 6- Photomicrograph of a cyst lined by ciliated columnar epithelium. Adjacent soft tissue shows mixed inflammation with presence of cholesterol clefts (Inset- cholesterol clefts with foreign body giant cells) (H & E stain, 40 X).

DISCUSSION

The branchial apparatus was first described by Von Baer. The origin and classification of different branchial anomalies (BA) is controversial and not clear even today. Many theories like thymopharyngeal duct theory, inclusion theory, degenerative cystic changes of cervical lymph nodes theory have been reported but most widely

accepted theory is the incomplete involution of branchial apparatus during embryogenesis.⁶ Out of six pairs of branchial apparatus, fifth arch usually disappears and the sixth arch is often represented as a part of fourth arch due to its small size.^{2,3} Incomplete involution of any of the four primary pairs of branchial pouches and branchial clefts give rise to branchial anomalies (BA) and they are classified according to their location as first, second, third and fourth arch anomalies.⁴

Table 1: Work's classification of first branchial cleft anomaly.

	Clinical presentation	Origin	Histopathological findings
Type I	Cystic mass	Ectoderm	Squamous epithelium
Type II	Cyst, fistula, sinus	Ectoderm and mesoderm	Squamous epithelium with cartilage or skin adnexa

Second branchial anomalies are the commonest accounting for 95% of cases, however first BA are seen only in 1-4% of case. ^{7,8} Third and fourth arch anomalies are extremely rare. ^{9,10} BA can present as cyst, fistula, sinuses and cartilage remnants in head and neck region, and they account for 20% of all pediatric congenital neck lesions. ^{1,11}

Branchial cysts typically present in older children, whereas fistulas are present in infants.¹² BA may be bilateral in 2-3% of cases where they are often familial and associated with other anomalies.¹³

Fistula and sinus usually present as discharging pits in infancy. They have a tendency for recurrent infections, so need prompt intervention and diagnosed much earlier than cysts. Cysts are often painless, compressible slow growing neck masses in young adults. Histologically, they contain a turbid yellow fluid containing cholesterol crystals and are lined by stratified squamous epithelium. ²

A cyst develops when a cleft remnant forms an epithelial lined space without any communication to mucosa or the skin. Incomplete obliteration of any of the cleft or pouch leads to the formation of a sinus, a blind tract which may communicate either with the mucosa or skin. On the other hand, fistula is formed when both cleft and pouch fails to involute and forms communication between skin and mucosa.¹

Although diagnosis of BA is usually clinical, physical examination and detailed history is essential for diagnosis. But in cases of cyst, radiological investigations like Ultrasonography and Computer tomography scanning provide accurate anatomical details about the site, extent, nature of lesion and its relationship with surrounding structures(parotid gland, facial nerve, major neck vessels). CT scan is reported to be more useful than MRI in evaluating branchial anomalies. Fistulogram can be performed to delineate the length and course of fistula / sinus, but it is not feasible in children every time to inject contrast in the tract without sedation. However, fistulograms may not be necessary if the tract has been clearly identified via other forms of non invasive imaging

modality or direct visualization, or by probing with lacrimal probe or umbilical catheter.

Preoperative/intraoperative fibreoptic laryngoscopy should be done in suspected pyriform sinus fistulas (third and fourth arch anomalies) to check for an internal opening.¹⁶

Surgery is the treatment of choice for BA.¹⁶ Patients presenting with acute infection should be managed initially with antibiotics and aspiration or drainage if required, followed by definitive surgery after 4-6 weeks.⁴ To prevent recurrent infections and due to rare possibility of malignant transformation, these lesions need complete surgical excision of the cyst and fistula/sinus tract encompassing the external opening.¹⁷

Complications following surgery include recurrence, wound infection, facial neve paralysis (specifically in patients requiring superficial parotidectomy for first arch anomalies) and vagal nerve paralysis (in cases of second branchial cleft cyst excision, third and fourth arch BA). ^{16,4}

First branchial cleft anomalies (FBCA)

They account for 1-4% of all cases of BA. FBCA are due to incomplete fusion of the ventral portion of the first and second arches. FBCA have close relationship with parotid gland and facial nerve, as migration of facial nerve, development of parotid gland and obliteration of the cleft all occurs at the same time. As cleft closure proceeds from ventral to dorsal, lesions occur more often near parotid and ear region than at hyoid region. 18

The annual incidence of FBCA has been reported as 1 per 1,000,000, and they occur more frequently in females (M: F ratio is 1:2). The anomaly begins on the floor of the external auditory canal either at the level of the bony-cartilaginous junction or in the cartilaginous portion, follows the seam between the mandibular and hyoid arches, and ends in the submandibular region. ²⁰

FBCA, they can present as cystic mass or discharging pit in the Pochet's triangle area which consists of the EAC,

the hyoid body and the mandibular angle and are more common on the left side. ^{19,21} The lesion is easily misdiagnosed and usually present repeatedly with swelling, pain and purulent discharge from the skin opening. Preauricular sinuses, epidermal cysts, dermoid cysts, mastoid abscess, postauricular lympadenopathy and parotid lesion all can mimic FBCA.

Triglia et al reported delay of 3.5 years in making the diagnosis and management of FBCA, i.e. average time taken between the onset of the first clinical symptom and when adequate treatment is received.²⁰ Contrast enhanced CT and magnetic resonance imaging help in exact localization of lesion and its relation with facial nerve, parotid gland and EAC.^{20,21}

Several authors have proposed classifications to assist appropriate diagnosis and management of these lesions. In 1971, Arnot classified FBCA into two types according to anatomical site: type I presents as a defect in the parotid region and seen in early or middle adult life, whereas type II defects are encountered during childhood in the anterior cervical region.²²

On the basis of clinical features and histology, Work divided FBCA into two types (Table 1).²³

In 1980, Olsen et al proposed a simplified classification into cysts, sinuses and fistulas.²⁴

Early diagnosis and treatment are needed to avoid recurrent infection and secondary development of fistulous tracts. Omplete surgical excision with wide exposure of the lesion is the only treatment for FBCA. The course of the tract varies and has variable relationship with the facial nerve-superficial, deep to the nerve or between the branches of the nerve. Thus, facial nerve is always at risk during the surgical removal of FBCA. The risk of facial nerve palsy is higher in patients having recurrent infections and inadequate treatment (incision, drainage or incomplete excision). Hence, it is advisable to perform a superficial parotidectomy in cases of FBCA while identifying the tract in relation to the facial nerve.

We also encountered collaural fistula in female child on left side, which is consistent with literature.

Second branchial arch anomalies

Second branchial arch anomalies were first described by Bailey in 1929.²⁷ Most of the second branchial cleft anomalies present within the submandibular space but they can occur anywhere along the course of the second branchial arch tract, which extends from the skin overlying the supraclavicular fossa, between the internal and external carotid arteries, to the oropharnx at the levels of tonsillar fossa.¹¹ Second arch anomalies may take several forms and most commonly present as cysts followed by sinuses and fistulae.¹⁴

A small opening or pit in the neck, near mid or lower part of sternocleidomastoid (SCM) muscle is most likely to represent a second branchial fistula/sinus.² As it ascends it pierces the platsyma. At the level of hyoid, tract passes between the external and internal carotids in relation to the hypoglossal and glossopharyngeal nerves, and open in to the oropharynx usually in the intratonsillar cleft of palatine tonsil.

A complete branchial fistula with external and internal opening is rare and most of the time external opening extends up the neck for a variable distance and ends blindly to form a sinus. Persistent mucoid discharge and/or salivary secretions from an opening in the skin of the neck, indicates complete fistula tract but dye test is used for confirmation, in which methylene blue is injected through the outer opening and appear in the throat. Sometimes the complete tract may be blocked by secretions or granulations and may give negative dye test. Histologically, sinus tracts of BA are typically lined by stratified squamous epithlelium and sometimes with respirator epithelium.

Bilateral fistulas of the first and second branchial arches are associated with genetic disorders and branchiootorenal (BOR) syndrome, thus complete head and neck and abdominal evaluation should be done in these cases.²⁹

In 1929, Bailey classified second branchial anomalies into four subtypes:²⁷

Type I- Most superficial and lies along the anterior surface of sternocleidomastoid deep to the platysma but not in contact with the carotid sheath.

Type II- this is most common type, where the branchial cleft cyst lies anterior to the sternocleidomastoid muscle, posterior to the submandibular gland, adjacent and lateral to the carotid sheath.

Type III- extends medially between the bifurcation of the internal and external carotid arteries, lateral to the pharyngeal wall

Type IV- It lies deep to the carotid sheath within the pharyngeal mucosa and opens into the pharynx.

Second branchial cleft cysts are usually painless and compressible lateral neck masses. They are filled with turbid yellow fluid containing cholesterol crystals and are lined by stratified squamous epithelium.²

A cyst in the lower anterior or lateral region of the neck should be differentiated from occult thyroid primary, malignant node (along the anterior border of SCM muscle). Therefore, ultrasonography, computer tomography and MRI are important tool for confirming the location, and nature of neck swellings. However, ultrasound and CT scans have no advantage in the visualization of the complete fistulous tract.³⁰ Fine needle aspiration is also mandatory before taking patient for any invasive procedure.

If not treated surgically, these lesions do not regress and may lead to recurrent infection. A step-ladder incision is described by Bailey in 1933 for complete exposure of the fistula tract. First incision is given at the sinus opening and second overlying the carotid bifurcation, tract traced till the tonsillar area and excised after ligation. However, Maddalozzo used single wide incision and ensured complete transection of the tract by ligating cephalad to the hyoid bone. Complete branchial fistula can be approached perorally after tonsillectomy.

For branchial cystic lesion, complete and meticulous dissection of the cyst from adjacent vital structures like carotid vessels, internal jugular vein (IJV), vagus nerve, hypoglossal nerve should be done.

Recurrence rates for surgical excision of branchial anomalies are as high as 22%.³¹ However, in 1996, Agaton–Bonilla and Gay-Escoda found reccurence in 8.6% cases.³² Thus complete surgical excision of cyst & fistula is the only treatment available.

Third and fourth branchial arch anomalies

These anomalies are very rare. 9,10 These appear similar to second branchial cleft anomalies, externally with a cutaneous opening in the supraclavicular area along the anterior border of SCM muscle, however internally they enter the pharynx through the pyriform sinus below the hyoid bone. 2

A third branchial arch anomaly originates from base of the pyriform sinus, passes through thyrohyoid membrane and is located above the superior laryngeal nerve. ³³ They course inferiorly, posterior to the internal carotid artery and anterior to vagus nerve, and end over the anterior border of the SCM muscle between middle and lower third as external cutaneous opening. ¹⁰

Theoretically, a fourth branchial cleft fistula/sinus tract arises from the apex of the priform sinus, which pierces cricothyroid membrane and is located beneath the superior laryngeal nerve. ³³ It then runs in the tracheoesophageal groove, behind thyroid gland and descends inferiorly to the mediastinum, looping around the arch of aorta in the left and subclavian artery in the right and ascends back into the neck, posterior to common carotid artery to reach the hypoglossal nerve, where a second loop is made around the nerve to end at the anterior border of SCM muscle. ^{29,34} According to Godin et al, all fourth arch anomalies occurred in the left side. ³⁵

These lesions present with repeated neck infections, thyroiditis and neck mass. It is therefore not easy to distinguish the third and fourth BA's in the surgical

operation on the basis of the tract and superior laryngeal nerve. 9,10

Wanpeng et al use collective term pyriform sinus fistula, for third and fourth BA, as they both share similar presentation and management. Several methods are used to identify the presence of internal openings of branchial anomalies. These include radiographic imaging (contrast swallow or fistulograms), or direct visualization by flexible fibreoptic scopes, an intraoperative laryngoscope or an oesophagoscope. However, these are simple and safe methods but usually not tolerated by young children. These lesions never regress, thus the treatment of choice is surgical removal of the fistula along with occlusion of the internal opening. Intraoperative use of methylene blue dye allows better localization of internal fistula.

Recently, endoscopic assisted cauterization of the internal opening has been used in the treatment of pyriform sinus fistulas.³⁶

Key message

- Single incision is sufficient for complete excision of neck fistula in children; hence stepladder incisor can be avoided, if operated in early age.
- Contrast study/fistulogram is not required especially in children, as diagnosis of branchial anomalies is usually clinical.
- We recommend imaging (MR/CT) for all cystic lesions.

CONCLUSION

Branchial anomalies (BA) are result of aberrant embryonic development of branchial arches. Complete history, examination and sound knowledge of anatomy are essential for clinical diagnosis. Complete surgical excision is the only permanent cure of BA.

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