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OTHER ARTICLES

Perplexing First Branchialcleft Anomalies—A Case Series with Review of Literature

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Abstract

Incomplete obliteration of the branchial apparatus results in the formation of branchial cleft anomalies. First branchial cleft anomalies may persist anywhere in the first branchial arch, from the external auditory canal at the level of the bony cartilaginous junction to the submandibular triangle. The majority of cases present in childhood as an opening in the skin though they may present as cysts or neck masses, mostly mistaken for neck abscesses which leads to inadequate treatment and complications. Here different cases of first branchial cleft anomalies with variable presentation and treatment are illustrated. The need for proper diagnosis and adequate treatment cannot be overemphasized to avoid mismanagement and complications.

Keywords Branchial cleft anomalies · Facial nerve · First branchial cleft anomalies · Frey's syndrome · Head and neck surgery · Pediatric otorhinolaryngology · Poncet' s triangle

Introduction

The branchial apparatus is the embryological precursor of the developing face, neck and pharynx from the fourth through seventh week of gestation. The most widely accepted theory for branchial cleft anomalies is the incomplete obliteration of branchial apparatus, commonly clefts and in case of fistulas and cysts the pharyngeal pouch also is implicated. They may have different presentation clinically and present as cysts, sinus tracts or fistulae [1]. Congenital branchial cleft anomalies are the second most common congenital lesions in the head and neck in children [2]. Second branchial cleft defects at 95% of these, are most common while first branchial cleft anomalies are reported to be less than 8% with third and fourth arch anomalies being extremely rare. [3, 4].

The other theories include persistence of vestiges of the precervical sinus, the thymo-pharyngeal theory and the cervical lymph node theory [5, 6]. Here different cases are presented with first branchial cleft anomalies with varying presentation, treatment and the emphasis is laid on the need for proper diagnosis and adequate management to avoid mismanagement and complications.

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Illustrative Cases

Case 1

A 24-year-old girl presented to the ENT OPD for recurrent left facial infection. Clinical examination revealed an unsightly scar around an orifice exuding pus located above the left angle of mandible. An opening was present in the left external auditory canal with pus. Sinogram showed a fistulous tract extending from the skin through left parotid region into a pocket of collection near the external auditory canal. MRI revealed thin-walled cystic lesion in deep lobe

of left parotid extending inferiorly superficially just above the angle of mandible (Figs. 1, 2 and 3).

Surgical excision was performed under general anaesthesia by the modified Blair incision. The facial nerve was identified and preserved. The fistula cord was dissected from its external skin opening and followed up through the deep lobe coursing medial to the facial nerve with a posterior cyst reaching up to the external auditory canal (cartilaginous part). Duplication of cartilaginous part of external auditory canal was present in the middle of the parotid tract. Facial nerve was dissected away and retracted with a nerve loop. It was dissected and excised along with cuff around opening in the floor of EAC. This fistula was Work type 2 first branchial cleft anomaly. No facial weakness was seen in post op period. The follow-up period was 12 months. The clinical examination did not reveal any sign of recurrence.

Case 2

An 8-year-old female patient presented with a post aural swelling on the right side of her neck with a history of sinus discharge and facial flushing and sweating on feeding. The cystic lesion was first noted months after birth, but it was smaller in size. She had incision and drainage from the site three times in primary health care centres. This led to excessive scarring of tissue and pre–operative Frey's syndrome (which might be due to injury to auriculotemporal nerve following multiple incision drainage and scarred tissue). CT Sinogram was done.

Surgery was the preferred choice of treatment. An elliptical incision was given around the sinus opening, tract

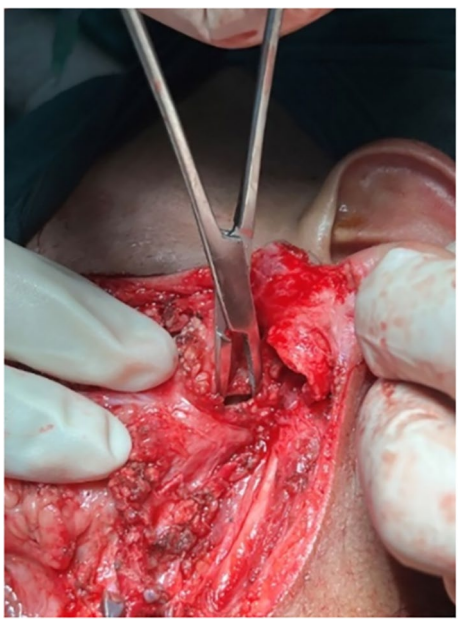

Fig. 1 Fistula tract medial to facial nerve branches

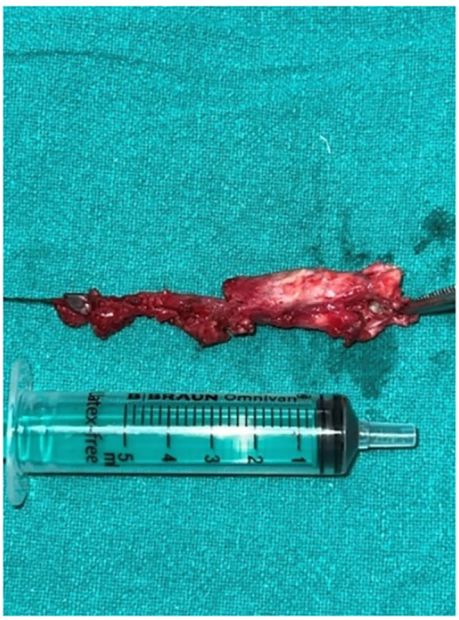

Fig. 2 Fistula tract and cyst; duplicated part of cartilaginous external auditory canal

was identified and excised up to the inferior cartilaginous part of external auditory canal. This was a work type 1 first branchial cleft anomaly. Post op was uneventful with no recurrence on follow up. However, symptoms of Frey's syndrome persisted in the post-op period. The patient was advised tympanic neurectomy but parents declined (Figs. 4 and 5).

Case 3

A 5-year-old female with an abscess in left post auricular region presented to ENT OPD. She had history of drainage from the site in primary centres and recurrence

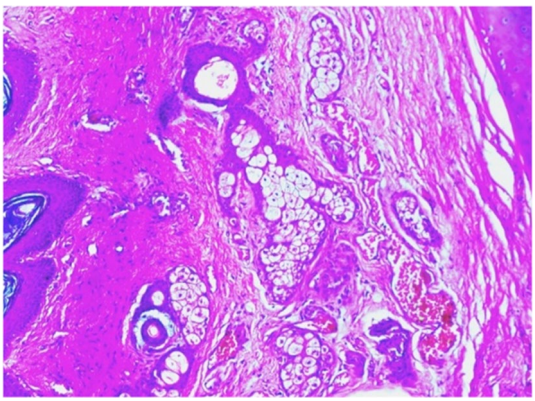

Fig. 3 Histo-pathological examination showed lining of epithelium with many pilo- sebaceous units and many adnexal structures. Hyaline cartilage and areas of fibrosis also seen

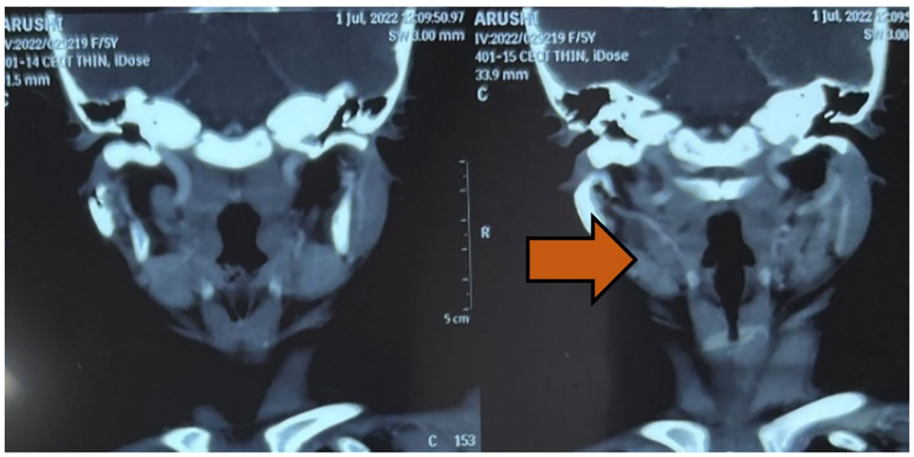

Fig. 4 CT scan coronal section: arrow pointing the fistula tract

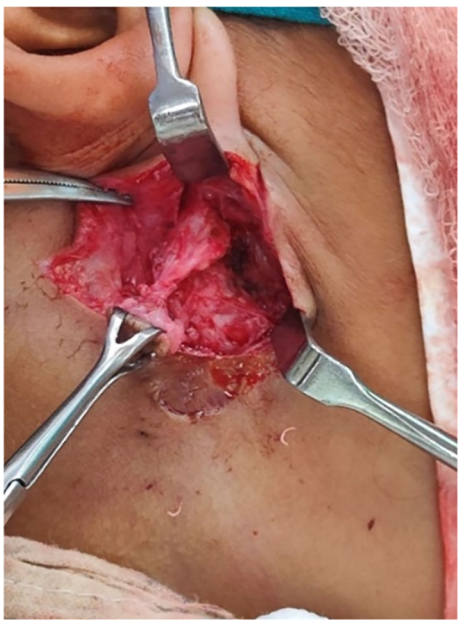

Fig. 5 Fistula tract. Unsightly scarring of skin can be seen because of multiple incision drainage which ultimately led to Frey's syndrome

every time. The surrounding skin was scarred. No opening was seen in external auditory canal and there was no history of aural discharge. The patient was taken up for surgery. An elliptical incision was given around the opening. The tract was identified, separated from surrounding tissue and was traced posteriorly up to cartilaginous part of external auditory canal and was excised in toto with cartilage cuff of external auditory canal. This was work type 1 first branchial cleft anomaly. Post op was uneventful and there was no recurrence on follow up (Fig. 6).

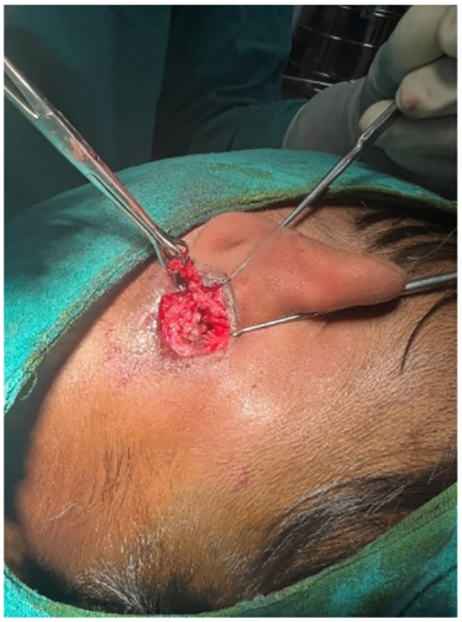

Fig. 6 Fistula tract extending from infra -auricular region to cartilaginous part of external auditory canal

Case 4

A 27-year-old male patient came with history of swelling and discharging sinus present in left infra-auricular region and multiple (two) openings in the pre -tragal region and inferior end of anti-helix. There was no opening in the external auditory canal. Past history of abscesses and incision and drainage several times was present. Patient was taken up for surgery. Tract was identified in the preauricular region and dissected up to the parotid fascia. A branch of tract reaching the pinna was also dissected and removed along with a cuff of auricular cartilage. This was work type 1 first branchial cleft anomaly. Post op was uneventful (Fig. 7).

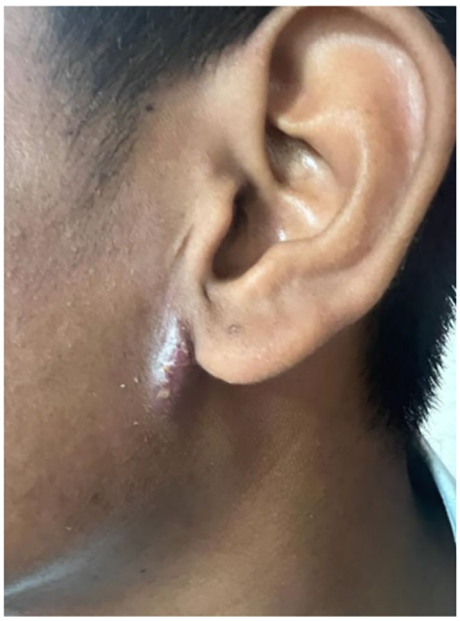

Fig. 7 clinical photograph showing swelling and discharging sinus in left infra-auricular region

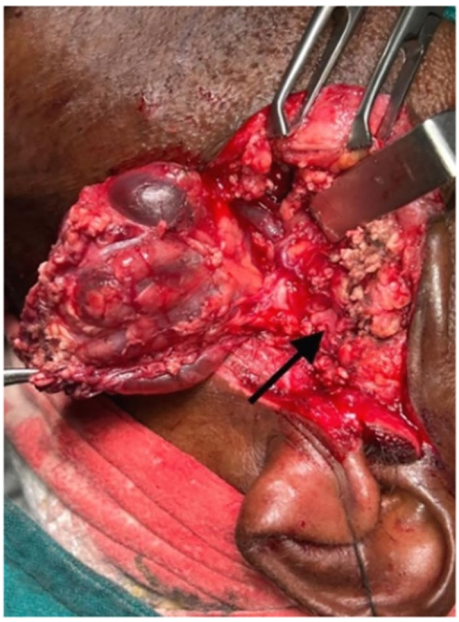

Fig. 8 Cystic lesion in left parotid region lateral to facial nerve branches

Case 5

A 65-year-old male patient presented with swelling near left side angle of mandible for 20 years and anterior neck swelling for 7 months. The patient never took any kind of treatment for this swelling as it was not increasing in size. Fine needle aspiration cytology from the facial swelling revealed a branchial cyst while the anterior neck swelling came as follicular neoplasia. CT scan revealed cystic lesion in left parotid region and bulky right lobe of thyroid.

Surgery was performed by Modified Blair incision. After identifying the facial nerve trunk first, the cyst was dissected and excised in toto lateral to the branches of facial nerve. No post op facial weakness was present. This was a type 2 branchial cyst (Figs. 8 and 9).

Review of Literature and Discussion

During the 4th week of embryogenesis, six pairs of branchial arches appear. During seventh week, the arches fuse and the clefts obliterate. Incomplete fusion of the ventral portion of the first and second arches leads to first branchial cleft anomalies [7]. The closure time of the cleft is simultaneous with the migration of the facial nerve and developing parotid gland, thus first branchial cleft anomalies have a close temporal relationship with these structures. Lesions occur more often near the ear and parotid gland area than at the hyoid region as clefts obliterates from ventral to dorsum [8].

In 1971, an anatomic classification was proposed by Arnot [9]. Here, Type I anomalies as defects in the parotid region, appearing during early or middle adult life and Type II as those that appear in the anterior cervical triangle with a communicating tract to the external auditory canal usually developing during childhood. In 1980, Olsen et al. proposed a simplified classification subdividing the defects as cysts, sinuses or fistulas. [10].

Karmody [11], further extended the concept of first branchial cleft anomalies including congenital anomalies of the external ear. He proposed four types of anomalies including aplasia, atresia, stenosis, and duplication. Classification

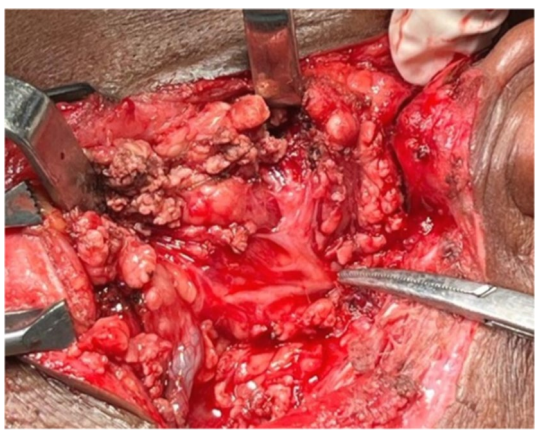

Fig. 9 Branches of facial nerve after removal of the branchial cyst

proposed by Karmody and Olsen et al. has been found to be clinically more relevant.

Work, in 1972 [12], described two types of first branchial cleft anomaly based on tissue of origin:

(1) Type I—These are of ectodermal origin. These are generally cysts, and are mostly duplication of the membranous external auditory canal. They present as periauricular cysts medial to concha, running parallel to external auditory canal and superior to facial nerve, may extend inferior or posterior to pinna. Histologically, they have a squamous epithelium lining and end in a cul de sac on a bony plate at the level of mesotympanum.
(2) Type II—These are mostly considered as duplication of cartilaginous and membranous part of external auditory canal. These may present as cysts or sinuses or fistulas of ectodermal and mesodermal origin. The most common presentation is a swelling in relation to parotid gland as an abscess around the angle of the mandible, with a track running up to the external auditory canal, which has a variable relationship with the facial nerve. Histologically, they are lined by squamous epithelium with cartilage and adnexal structures.

J. Wilson et al. proposed a revised classification method for first branchial cleft anomalies based on the absence (type I) or presence (type II) of parotid gland involvement. [13].

Fistulas are often closely associated with the facial nerve, which is prone to damage during the operation, thus causing facial paralysis postoperatively and affecting the quality of life of children. Solares et al. [14] studied 10 patients with branchial cleft anomalies and stated that 7 out of 10 lesions ran medial to the facial nerve, 2 were lateral and 1 ran between the branches. Triglia JM et al. in a review of 73 cases showed that the anomaly passed medial to the nerve in 21 cases (29%) and was split around the nerve in 6 (8%) [15]. Fistulas have a tendency to run deep into the nerve whereas sinus tracts tend to run lateral to it.

The most common presentation is swelling in the periauricular region (24%) [16]. Anatomically first branchial cleft cysts or their sinus orifices are located in Poncet's triangle. Poncet's triangle [17] is bounded superiorly by external auditory canal, anteriorly by mental region and inferiorly by hyoid bone, lying anterior to sternocleidomastoid muscle (Fig. 10).

Anatomically first branchial cleft cysts or their sinus orifice are located in this triangle. The common sites of external opening of fistulas and sinuses are external auditory canal (40%), upper neck (32.5%), concha (20%), and post-auricular area (7.5%) [18]. Uncommon sites include middle ear, parapharyngeal space [19] and Eustachian tube [20, 21]. The external auditory canal may reveal the sinus, fistula opening as a pit in the floor. There may however be complete absence of signs in the external auditory canal. The periauricular swelling may push the walls of external auditory canal to partially or totally obstruct the canal. Sichel et al. [22] stated that type II first branchial cleft anomalies were associated with a myringeal web, an epidermal web extending from floor of external auditory canal to umbo or pars tensa of the tympanic membrane. Usually, tympanic membrane and middle ear are normal.

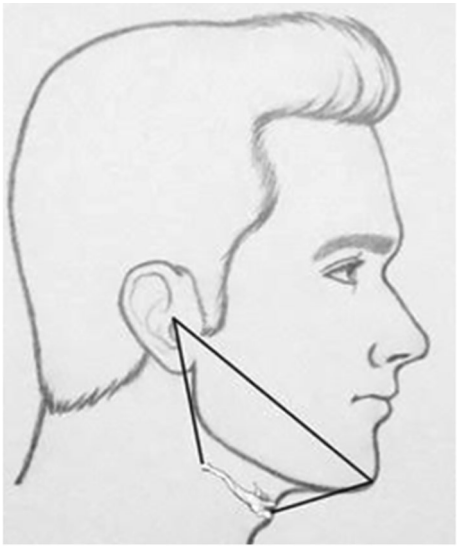

Fig. 10 Poncet's triangle

Yalcin et al. [23] reported a case where a first branchial cleft lesion was associated with cholesteatoma of mastoid and middle ear, microtia and aural atresia. Bilateral ear involvement has also been reported [24].

In case 2, the patient presented with pre -operative Frey's syndrome. This syndrome has never been reported before in the world literature in relation to first branchial cleft anomalies. Frey's syndrome or gustatory sweating usually occurs with auriculotemporal nerve injury as a result of surgical intervention or trauma in the parotid region which leads to aberrant reinnervation of the auriculotemporal nerve [25]].

The facial nerve and the first branchial cleft are closely related more than in any other type of branchial defect. So, the main aim during surgery is preserving the facial nerve and excising the entire tract. The first branchial anomaly presents in an area that is cosmetically significant and an unplanned approach may lead to embarrassing complications. Early diagnosis and treatment is the cornerstone of these anomalies.

Conclusion

The first branchial cleft fistula being a rare entity, it is mostly misdiagnosed and mistreated. They should be suspected in patients with otorrhea, in the absence of chronic otitis, an

orifice located in the area of the neck defined by the hyoid bone below, the sternocleidomastoid muscle in the back, and the edge of the mandible in the front communicating with the external auditory canal and in patients with isolated cysts in the parotid gland that increase in size during an inflammatory period.

Early diagnosis and proper treatment are imperative for avoiding recurrences and preventing damage to the facial nerve [5]. Therefore, the surgical treatment is challenging. CT and fluoroscopic studies can be instrumental in minimizing surgical complications by establishing a definitive diagnosis and directing treatment [24]. Dissection and complete removal of the sinus or fistulas is less exigent in absence of any previous infection and surgery can be safely performed at any time after the 6 months of age. A high index of suspicion among the general physician, otorhinolaryngologists, paediatrician and dermatologists is important for making a right and informed diagnosis and to prevent complications like damage to facial nerve and auriculotemporal nerve through inadvertently exploring the lesion.

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Declarations

Conflict of interest I declare that we have had no relevant financial interests or personal affiliation / no conflict of interest. declare that all authors have agreed to submit the manuscript to the journal. (Indian Journal of Otolaryngology and Head & Neck Surgery).

Consent for publication I declare that this work is original and the manuscript is not currently under consideration for publication elsewhere

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