Case Report

First Branchial Cleft Anomalies have Relevance in Otology and More

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Abstract

Introduction: First branchial cleft anomalies account for less than 8% of all branchial abnormalities. Their rarity and diverse presentations have frequently led to misdiagnosis and inappropriate treatment. In a trend towards specialisation/subspecialisation, first branchial cleft duplication anomalies, with their varied clinical manifestations, may possibly present to an Otology, Head and Neck Surgery, Paediatric Otolaryngology, Maxillofacial or even a General Paediatric and General Surgery practice. There is a need to highlight the clinical features which can aid in accurate diagnosis. Clinical Picture, Treatment and Outcome: A case of an adult with Work Type 2 first branchial cleft duplication anomaly presenting as a collaural fistula is described. It first presented as a recurrent upper neck abscess in childhood. The diagnosis had previously been missed although the patient was able to clearly establish a correlation between digging of the ipsilateral ear and precipitation of the abscess. Instead of an epidermal web, a myringeal lesion in the form of a fibrous band-like was present. The lesion was completely excised with no further recurrence. Conclusion: This case highlights useful diagnostic features both from the history and physical examination. The specialist/subspecialist must be aware of this condition and be mindful of its possible cross specialty/subspecialty symptoms and signs. Together with a good understanding of the regional embryology and anatomy, the lesion can be diagnosed early at initial presentation with the potential for best treatment outcomes.

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Introduction

The external ear canal is a derivative of the first branchial cleft. Anomalies of the first branchial cleft, therefore, often involve external ear canal structures which are normally managed in an Otology practice. However, in duplication anomalies, clinical features are varied and may include those seen in a typical Head and Neck Surgery, Paediatric Otolaryngology, Maxillofacial or even a General Paediatric and General Surgery practice. Because of its rarity and diverse cross specialty/subspecialty clinical manifestations, the condition is often overlooked and mismanaged, which can result in dire consequences. This paper reports a case to highlight specific diagnostic clinical features and discusses the significance of this condition in Otolaryngology.

Case Report

A 48-year-old man was referred to us for further management of a recurrent discharging left upper neck abscess. The abscess first appeared when he was still a young boy and since then, he had had numerous incision and drainage procedures performed for the recurrent infections. Typically, a neck abscess developed shortly each time he dug his left ear. He was otherwise healthy.

On examination, there was a small cutaneous opening in the left submandibular region (Fig. 1). The area surrounding the skin opening was scarred due to previous infections and surgeries. A small fistulous opening was also noted at the floor of the cartilaginous portion of the left external auditory canal. The left eardrum showed a thick band of tissue (fibrous in appearance) extending from the umbo to the floor of the ear canal (Fig. 2). Computed tomography (CT) scans showed a wide fistulous tract connecting the cartilaginous portion of the left external ear canal to the neck and traversing the deeper aspect of the parotid (Fig. 3). A diagnosis of first branchial cleft fistula was made.

During surgical exploration, the main branch of the facial nerve was identified and traced distally. The fistulous tract

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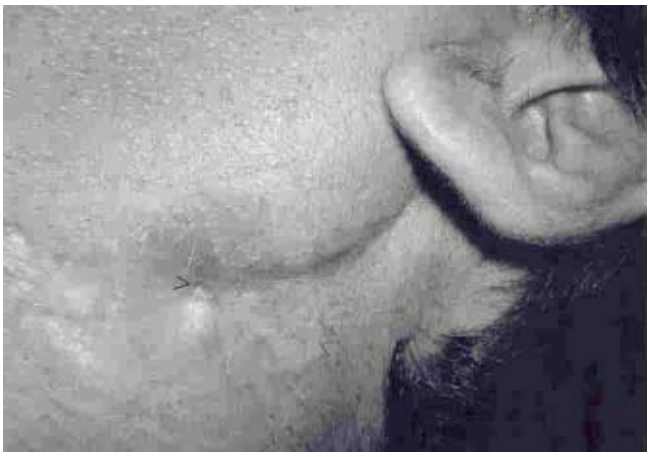

Fig. 1. The cutaneous fistula opening (indicated by arrow-head) within the left Pochet's triangle (bounded by the external auditory canal superiorly, mental region anteriorly and hyoid bone inferiorly). The surrounding tissues are scarred from previous infections and surgeries.

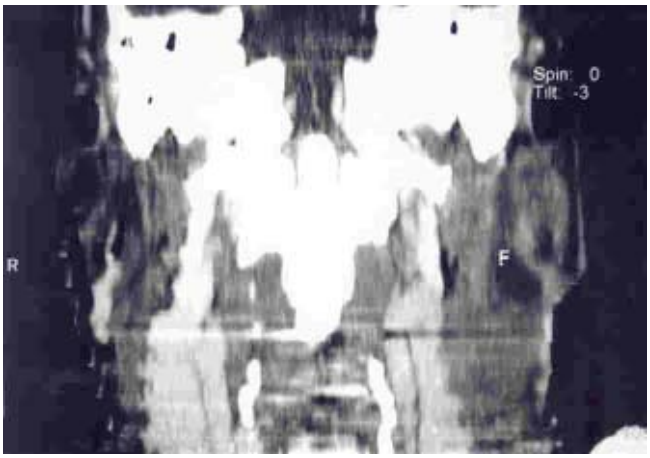

Fig. 3. Reconstructed coronal CT scan view showing a wide fistulous tract (F) connecting the cartilaginous portion of the left external auditory canal to the upper part of the neck. Lateral to the fistula tract is an associated parotid cystic cavity.

was noted to run medial to the main trunk of the facial nerve. The aural end of the tract was thick and lined with cartilage, while the cervical end was enmeshed in inflamed fibrous tissue. The tract was completely excised with a small cuff of external ear canal cartilage and skin, with primary closure. The main branches of the facial nerve could be preserved, except for the cervical branch which was embedded in the inflammatory mass of fibrotic tissue. The postoperative recovery was uneventful.

Histological examination of the excised fistula confirmed that it was lined by squamous epithelium with adnexal structures.

Incidence

First branchial cleft anomalies are rare, with only about 200 cases reported in the literature.[1] It accounts for less than 8% of all branchial abnormalities.[2,3]

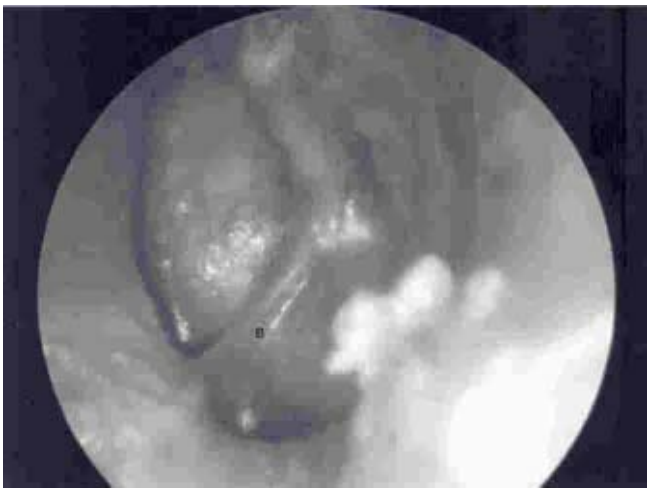

Fig. 2. Instead of a epidermal web-like myringeal structure, the left tympanic membrane demonstrated a fibrous band-like tissue (B) connecting the umbo to the floor of the external auditory canal.

Embryology

During the 4th week of human embryological development, 6 pairs of branchial arches appear which will form the future lower face and neck;[3] and they disappear by the 7th week. Mesodermal in origin, these arches are separated from each other by the 5 branchial clefts (ectoderm) externally and 5 pharyngeal pouches internally (endoderm). First branchial cleft anomalies are a result of incomplete closure of the cleft.[4] The chance of malformations occurring nearer the ear and parotid is greater than that occurring at the hyoid region, as obliteration of the cleft proceeds from ventral to dorsal.[5] Although the lesion normally has a close relationship to the parotid and facial nerve, the relationship is variable, presumably because of temporal differences during development.[4]

Classification

In 1971, an anatomic classification by Arnot[6] designated Type 1 anomalies as defects in the parotid region, appearing during early or middle adult life. Type 2 defects appear in the anterior cervical triangle with a communicating tract to the external auditory canal and usually develop during childhood.

In 1972, Work[7] proposed a histological classification. Type 1 anomaly is a defect of ectodermal origin, arising from duplication of the membranous external auditory canal. Clinically, they appear as soft cysts lined by squamous epithelium. It can have a tract running medial and parallel to the external auditory canal, superior to the facial nerve and ending in a cul-de-sac on a bony plate at the level of the

mesotympanum. The overlying skin is normal but accidental rupture or secondary infection may result in an intra-meatal or a retroinfra-auricular sinus opening. Type 2 defects are ectodermal and mesodermal in origin, containing skin with adnexal structures as well as cartilage. They present as a cyst, sinus, fistula or a combination.[8] They are associated with a sinus/fistula opening in the region of the submental triangle, extend superiorly through the parotid gland towards the floor of the external auditory canal at the level of the bony-cartilaginous junction or the cartilaginous portion.

In a third classification, Olsen et al[9] in 1980 classified the defects as cyst, sinuses or fistulas.

Relevance in Otology

Although congenital in origin, first branchial cleft anomalies can present later in life, at a mean age of 18.9 years (range, 20 days to 82 years).[2]

Otorrhea is the most frequent otological symptom and the condition should be suspected if recurrent/chronic otorrhea is present in the absence of chronic otitis. A sinus/fistula opening in the external auditory canal is present in only 44% of patients;[5] and even if such an opening exists, it may not necessarily appear obvious.[10]

In 2 out of the 3 cases reported by Sichel et al,[10] first branchial cleft anomalies are associated with a myringeal web, an epidermal structure which extends from the floor of the external auditory canal to the umbo of the tympanic membrane. However, in a larger series reported by Triglia et al,[5] it was found in only 10% of the patients. Although it is generally accepted that a direct connection between the fistula/sinus tract and the web does not exist, the tympanic membrane and middle ear are rarely involved by the tract.[11] Sinuses and fistulas arise from incomplete closure of the first branchial groove and if the failure occurs during the time of formation of the closing membrane, a fistula extending into the middle ear (a first pouch derivative) may develop.

First branchial cleft anomalies can also be associated with other otological conditions. Yalcin et al[8] reported a case where a first branchial cleft lesion was complicated by both cholesteatoma and aural atresia.

A Type 1 cyst can be removed while still keeping the epidermal skin of the meatus intact via a retroauricular incision.[3] Work recommended marsupialisation of this cyst through the external ear canal.[7] For a Type 2 lesion, early identification of the facial nerve at the stylomastoid foramen is recommended.[3] If this part is affected by disease, identifying the facial nerve proximally in temporal bone and tracing it distally may be the safest option.[12] Should the sinus/fistula opening involve the external auditory canal, it is removed with skin and cartilage. Primary closure is normally possible but if more than 30% of the circumference of the external auditory canal is denuded, split-thickness skin grafting and stenting are recommended.[13] If the tympanic membrane or middle ear structures are involved, reconstructive otologic surgery may be necessary.[11]

Relevance in Head and Neck Surgery

Accurate diagnosis of first branchial cysts located in and around the parotid gland can be difficult without surgical exploration.[5] Poncet's triangle is the anatomical triangle where first branchial cleft cysts or their sinus orifice are typically located.[10] The limits of the triangle are the external auditory canal above, the mental region anteriorly and the hyoid bone inferiorly.

The relationship of the lesion to the facial nerve is variable. In a series of 10 patients with first branchial cleft anomalies reported by Solares et al,[4] 7 lesions ran medial to the facial nerve, 2 were lateral and 1 ran in between the branches of the nerve. Fistulas have the tendency to run deep into the nerve whereas sinus tracts tend to run lateral to it.[2,4] Because of its variable relationship with the nerve, its removal warrants early identification and wide exposure of the nerve,[3,4] and/or the use of facial nerve monitoring.[13]

Relevance in Paediatric Otolaryngology

Type 2 first branchial cleft cysts often present in young children.[10] In the series reported by Stokroos and Manni,[3] 8 out of 18 patients presented at less than 10 years of age. Agenesis of the parotid can be associated and is suspected if a dimple anterior to the tragus is present,[14] and this can be confirmed by CT scans.[10]

In young children, the facial nerve lies more superficially and surgical landmarks used for identifying the facial nerve in adults may not be applicable. This necessitates the modification of surgical techniques to achieve safe and complete excision of the lesion.[14]

Management

Misdiagnosis is common in first brancial cleft duplication anomalies. Triglia et al[5] noted a delay of 3.5 years between the time of initial presentation and that when adequate treatment was received, with almost 50% of patients having a history of unsuccessful treatment.

The clinical picture of first branchial cleft anomalies usually overlaps with those seen in other more common diseases. This, together with its rarity, makes misdiagnosis common, leading to inadequate and inappropriate treatment. The resultant recurrent infections and repeated surgery lead to increased scarring and higher risk of iatrogenic facial palsy during its subsequent surgical removal. Furthermore, if unidentified, the incision and drainage of such an abscess may result in facial nerve injury.

In a trend towards specialisation and subspecialisation, a

patient with a first branchial cleft anomaly may present to an Otologic, Head and Head, Paediatric Otolaryngological, Maxillofacial or even a General Paediatric or General Surgical practice. The condition can easily be overlooked if the clinician adopts a compartmentalised specialty/subspecialist approach and fails to see the full clinical picture. Accurate diagnosis is key to successful treatment, especially at primary presentation.

Unlike some other branchial anomalies, first branchial cleft defects are rarely associated with other facial malformations which can contribute to making the correct diagnosis.[2] However, the condition is usually associated with infection[5] and therefore an inflammatory process in the region of the Pochet's triangle should immediately raise an index of suspicion. The usefulness of various anatomical and histological classifications is limited[13] and it is often difficult and confusing trying to correlate the clinical picture with the various classifications.[4,8] Analysis of clinical manifestations (cervical, parotid and auricular) and the findings of careful physical examination focusing on the external auditory canal are considered to be more helpful in achieving early diagnosis;[5] although fistulous openings into the external auditory canal and myringeal webs are not always present.

Besides the physical examination of the ear, this case highlights an additional diagnostic feature in that the patient could clearly establish a correlation between digging the ear and precipitation of an ipsilateral upper neck infection. Besides such a history of an aurogenic upper neck abscess, this case also illustrates the diagnostic significance of an associated myringeal lesion in this condition. However, instead of an epidermal web-like structure commonly described in the literature, it can be in the form of a fibrous band-like structure.

Imaging studies are useful in aiding diagnosis. CT scan can confirm the diagnosis by showing the tract near the external auditory canal to be wide and surrounded by cartilage.[10] A fistulogram is a useful diagnostic tool in the case of a sinus or fistula without signs of cyst formation or inflammation.[3]

The aim of treatment is to remove the lesion completely and safely with preservation of the facial nerve and to reconstruct otological structures if necessary. The best chance of achieving this is through early diagnosis at primary presentation and the availability of the necessary surgical expertise.

Conclusion

Although the external ear canal is derived from the first branchial cleft, the clinical manifestations of its duplication anomalies are not necessarily confined to Otology. Its rarity and diverse presentations have frequently led to misdiagnosis and inadequate treatment. The clinician must be familiar with its specific Otological as well as Head and Neck clinical features, in order to arrive at an early and accurate diagnosis. Surgical expertise must be available to remove the lesion completely, preserve all branches of the facial nerve if possible, and perform otologic reconstructive surgery if necessary.

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