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Main Article

Presentation of first branchial cleft anomalies: the Sheffield experience

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Abstract

Abnormalities of the first branchial cleft are rare. They may present with a cutaneous defect in the neck, parotid region, external auditory meatus or peri-auricular area, or with inflammatory or infective lesions at these sites.

A retrospective case note review of the patients treated by the senior author is presented. This group consisted of 18 patients and represents the largest published UK series to date. Eleven patients (65 per cent) had undergone incomplete surgery prior to referral.

Over half the patients had a clinically apparent lesion in relation to the external auditory meatus. There was a variable relationship between the tract and the facial nerve, which was identified at surgery in 15 cases.

These findings are consistent with those of previously published series. Clinicians should keep this diagnosis in mind when assessing patients with infected lesions in the neck and parotid area. Surgeons should be familiar with parotid surgery, in children where appropriate, and be prepared to expose the facial nerve before embarking on the surgical management of these lesions.

Key words: Branchial Clefts; Infant, Newborn Diseases; Head and Neck

Introduction

Congenital anomalies of the first branchial cleft are uncommon, accounting for fewer than 10 per cent of all branchial cleft anomalies.[1,2] Arndal *et al.*[3] estimate an annual incidence of one case per 1 000 000 population. The management of these rare lesions is challenging and requires knowledge of the embryological anatomy of the branchial apparatus.

The branchial apparatus appears between the fourth and fifth weeks of fetal development. It consists of six paired branchial arches separated by branchial clefts externally and branchial pouches internally. The fifth arch disappears early in fetal life. During normal development, the ventral portions of the first and second arches fuse, resulting in the disappearance of the ventral portion of the first cleft. The remainder of the first cleft forms the cavum conchae and the external auditory meatus, while the first branchial pouch forms the eustachian tube and tympanic cavity.

A number of authors have suggested classification systems for these anomalies. Arnot, in 1971,[4] described two anatomical types: type one, presenting with a defect in the parotid region, usually in early to mid-adulthood; and type two, presenting in childhood with a defect in the anterior triangle of the neck and a tract communicating with the external auditory meatus. This author proposed that type one defects resulted from cell rests being buried during the closure of the ventral portion of the first cleft, whereas type two defects arose from incomplete closure of the cleft.

Work, in 1972,[5] described two similar categories on the basis of histology and proposed embryology. Type one lesions are of ectodermal origin and occur medial to the concha, often extending into the post-auricular crease. Histologically, they show cyst formation and keratin, consistent with their ectodermal origins. Type two lesions are of ecto- and mesodermal origin and therefore contact skin and structures derived from mesoderm such as cartilage. The inferior opening of these lesions is below the angle of the mandible. They extend upwards, passing superficial to, deep to or through the branches of the facial nerve, to end in or around the external auditory meatus. In an unusual case described by Nichollas *et al.*,[6] a type two branchial cleft anomaly was associated with a cholesteatoma.

Karmody[7] included these lesions in a classification of all abnormalities of the external auditory meatus, dividing these into aplasia, atresia, stenosis and duplication. Olsen[1] reported a case series of 38 patients and simply divided these lesions into cysts, sinuses and fistulae. This last classification may be more clinically applicable as it provides practical

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information. Fistulae are said to usually be deep to the facial nerve, while sinuses and cysts are usually superficial to it.[8,9] A number of authors have observed that these lesions may be considered as duplications of the external auditory meatus.[10]

In a retrospective review of 39 cases, Triglia *et al*.[11] reported the results of surgical treatment. Of their patients, 17 (44 per cent) had undergone previous incomplete surgery, one of these cases being complicated by a facial palsy. Facial nerve dissection was required in 36 cases. In three patients, recurrence was observed requiring a second surgical procedure. The authors stressed the importance of early correct diagnosis.

In a more recent case series, including all fistulae and cysts of the neck, the same department classified first branchial cleft anomalies into three groups according to presentation: cervical, parotid and auricular.[12]

Leu and Chang[13] described two cases treated surgically without complication, apart from two recurrences occurring in the same patient.

In the largest published UK series to date, Ford *et al*.[2] described five cases of first arch anomalies within a study of 106 patients with other branchial cleft and pouch anomalies. These authors reported frequent delays in diagnosis and stressed the importance of early correct diagnosis, surgical treatment, and the need for facial nerve exposure and protection.

The senior author (PDB) has received referrals from Sheffield and surrounding regions as well as tertiary referrals from throughout the UK. The aim of this study was to determine, in this referral base, the modes of presentation, possible reasons for diagnostic delay, the anatomy of the defect (including its relationship to the facial nerve where appropriate) and the results of surgical treatment.

Methods

This was a retrospective case note review. The case notes of patients with first branchial cleft anomalies presenting to the Sheffield Children's Hospital and to the Royal Hallamshire Hospital and elsewhere from 1984 to 2006 were scrutinised. The following information was extracted: the age and sex of the patient; the site of the lesion; the mode and source of referral; any delay in diagnosis, referral or treatment; and the surgical treatment necessary, including the relation of the lesion to the facial nerve and histological findings. The eventual results, including any complications, were also noted.

Results

Eighteen patients were identified. The clinical data are summarised in Table I. Six patients were male and 12 were female. The mean age at presentation was 4.23 years (four years three months), with a range from birth to 24 years. Age at the onset of symptoms ranged from birth to nine and a half years. There were no bilateral lesions. One patient had atresia of the external auditory meatus on the affected side. No other patients had any clinically apparent congenital abnormalities of the head or neck.

The initial clinical manifestation was recurrent infections and/or discharge, in association with lesions, either of the neck or parotid (10 patients) or of the auricular or pre-auricular region (eight patients).

Eleven patients had undergone one or more previous surgical procedures, most commonly incision and drainage of neck abscesses (nine patients, four on two occasions). One patient had undergone excision of peri-auricular cysts and sinuses on four occasions prior to referral.

Eleven patients had defects in the anterior neck or parotid region (Figure 1). The remaining seven patients presented with lesions or infections in the ear or peri-auricular region. Where there was a lesion in the neck or parotid region, this was exposed using a parotidectomy approach and the tract dissected after exposure and dissection of the facial nerve.

In 14 cases, the tract was attached to the external auditory meatus, tragus or tympanic membrane. In three patients, the tract ended in a strand of fibrous tissue which crossed the external auditory meatus and ended at the umbo (Figure 2). One patient had a complete duplication of the ear canal which extended to the central aspect of the tympanic membrane. In twelve of our cases, the proximal end of the tract was clinically apparent in or near the ear prior to surgery, and in eight cases it had caused some infection or discharge at this site.

The facial nerve was identified in 15 of the 18 patients via a superficial parotidectomy approach (Figure 3). The tract ran deep to the facial nerve in eight cases, superficial to it in five, and passed between the branches of the nerve in two (in these two patients the tract and the nerve were adherent to each other).

It was not necessary to identify the nerve in three patients. In the first patient, the tract was followed from the conchal bowl (where it had been clinically apparent as a focus of recurrent infection) into the tissue of the parotid gland, where it ended. In this patient, the nerve was not identified but the tract was thought to be superficial to the nerve. In the second patient, the tract was followed from a pit on the cheek into the substance of the parotid tissue, where it ended. The third patient had a superficial tract extending from a punctum antero-inferiorly to the ear lobe to a cyst in the external auditory meatus. Deep to the cyst, the tract extended as a complete duplication of the ear canal to the central aspect of the tympanic membrane. The canal was repaired and packed after excision of the tract.

Histological information was available for 10 patients. All specimens were reported as tracts lined with keratinised stratified squamous epithelium, with associated cartilage and chronic inflammation. The senior author did not routinely request sinograms for these patients.

Complications included some persistent pain in one patient's post-auricular scar. This settled

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TABLE I

PATIENTS' CLINICAL DATA

Patient	Age at presentation	Presentation	Previous surgery	Tract	Relationship to facial n
1	2 y 11 m	Recurrent infection of neck lesion, discharging sinus	×2 I&D	Anterior neck to tragus	Superficial
2	9 y 6 m	Infected lesion in pinna	No	Conchal bowl to parotid tissue, passing through tragal cartilage	Not explored
3	2 y 10 m	Infected parotid lesion	I&D	Parotid to EAM	Deep
4	24 y 10 m	Infected post-auricular lesion	×4 explorations	Post-auricular area to tragus	Superficial
5	2 y	Infected post-auricular lesion and 'otitis externa'	No	Post-auricular area to EAM	Deep
6	1 y 2 m	Discharging facial pits	No	Into parotid tissue	Not explored
7	2 y 1 m	Infected parotid lesion	I&D	Parotid to EAM	Deep
8	4 m	Infected lesion in EAM	No	Parotid to EAM	Superficial
9	Birth	Discharging sinus in neck skin	No	Neck skin to EAM	Deep
10	2 y 3 m	Parotid mass	Exploration of mass	Parotid to tympanic membrane	Between branches
11	1 y 11 m	Recurrent abscess	I&D	Parotid to EAM (atretic EAM)	Deep
12	4 y 2 m	Parotid sinus	I&D	Parotid to mandible	Deep
13	10 m	Dimple in cheek, discharge & swollen right EAC	No	Cheek to tympanic membrane	Deep
14	1 y 7 m	Recurrent ear & cheek infection	I&D	Angle of mandible to EAM	Between branches
15	10 y	Infected haematoma in pre-auricular area	Cyst excision & ×2 I&D	Bifurcation of CN VII to TM	Superficial, and cyst at bifurcation
16	2 y 3 m	Recurrent abscess	×2 I&D	Parotid to EAM (duplicate EAC)	Deep
17	5 y	Recurrent abscess & discharging sinus below ear	×2 I&D	Neck to tympanic membrane (duplicate EAC)	Superficial and very adherent
18	7 m	Cystic lesion at EAM and occasional otorrhoea	No	Angle of mandible to tympanic membrane (duplicate EAC)	Not explored

n = nerve; y = year; m = month; I&D = incision and drainage; EAM = external auditory meatus; EAC = external auditory canal; CN = cranial nerve; TM = tympanic membrane

completely eight months post-operatively. One patient had weakness of the mandibular branch of the facial nerve pre-operatively which persisted after the operation. Another developed transient weakness of the nerve, which recovered completely at three weeks. Finally, one patient had a sinus in the conchal bowl which settled spontaneously and had not recurred at 30 months.

Discussion

During normal embryological development, the first branchial cleft forms the external auditory meatus. Anomalies of its development are caused by incomplete or anomalous closure of its ectodermal portion. The nature of the abnormality, whether it be a cyst, a sinus or a fistula, depends on the degree of its partial closure. These abnormalities may be considered as abnormal duplications of the external auditory meatus.[14] In common with previously reported case series, the tracts in the majority of our patients (83.3 per cent) involved the cartilaginous external ear, supporting this view.

Triglia *et al.*[11] observed that all their patients' cutaneous defects occured within a triangle with its apex at the external auditory meatus, its base a line between the tip of the chin and the middle of the hyoid bone, and its two remaining sides curving from the external auditory meatus to the tip of the chin along the body of the mandible and from the external auditory meatus to the greater cornu of the hyoid bone. They made the point that no lesions were observed to open below the level of the hyoid. The patients in our series confirm these observations. Many of their lesions opened in close relation to the parotid gland, and intra-parotid cysts may be caused by this mechanism. These lesions may be confused with parotid masses, especially when complicated by infection.

In common with other reported case series, we observed no bilateral lesions. One of our patients had an atretic external auditory meatus on the side of the lesion. Given the role the first cleft plays in the formation of the external auditory meatus, this association is not surprising.

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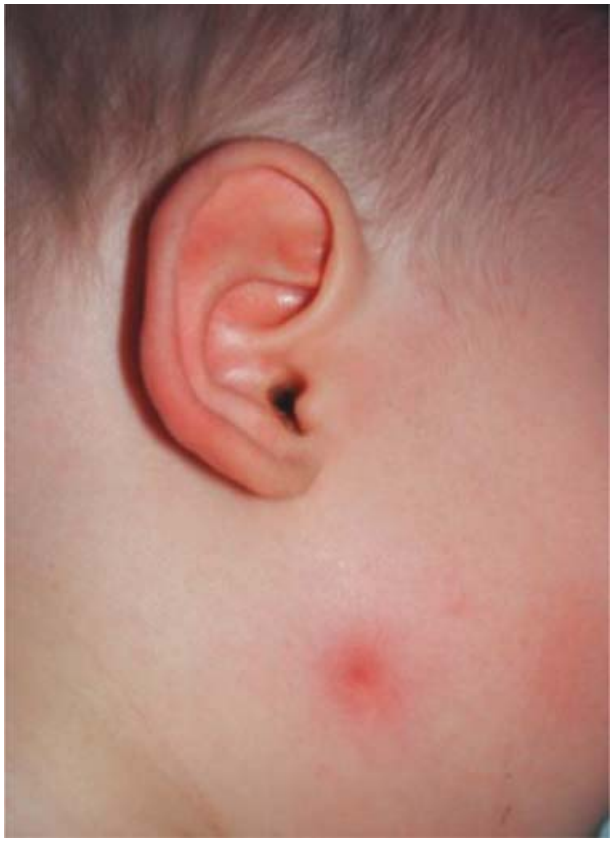

FIG. 1
First branchial cleft sinus presenting as a discharging sinus on the face.

Previous authors[1,4,6,8,9,11] have noted the connection of the tract to the external auditory meatus and associated structures. Three of our patients had a strand of fibrous material extending from the floor of the external auditory meatus to the umbo, and one had a duplication of the ear canal attached to the tympanic membrane. Similar abnormal connections between the external auditory meatus and the tympanic membrane were noted in four cases in the series reported by Triglia *et al.*[11] Olsen *et al.*[1] reported one case in which there was communication with the middle-ear space. This was not the case in any of our patients and is extremely rare, with only a few isolated cases reported.[15]

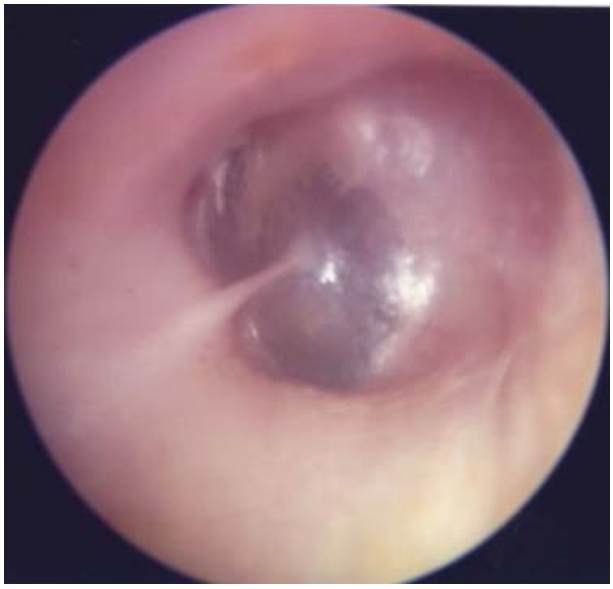

FIG. 2
First branchial cleft fistula extending from the canal wall to the umbo of the tympanic membrane.

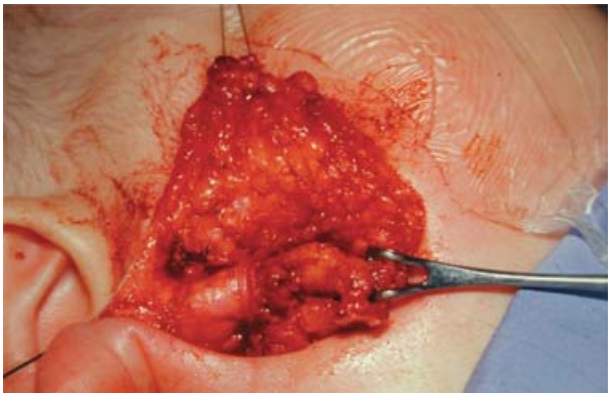

FIG. 3
First branchial cleft fistula, identified through a superficial parotidectomy approach and lying deep to the facial nerve (same case as shown in Figure 1).

There is a variable relationship between the facial nerve and the tract. In our series, eight tracts ran deep to the nerve, seven ran superficial and two ran between its branches. This finding is consistent with the observations of others. Triglia *et al.*[11] reported 14 out of 36 cases with tracts running deep to the nerve, 18 with superficial tracts and four with tracts running between the nerve's branches. Our case series does not follow the premise that fistulae run deep to the facial nerve and cysts and sinuses run superficial.[8,9] In our series, five (62.5 per cent) of the tracts that ran deep to the nerve were sinuses, and three of the seven tracts that ran superficial to the nerve were fistulae (43 per cent). Awareness of this variability, and a readiness to expose the nerve, is essential when undertaking surgical treatment of these lesions. Surgeons managing these patients should be familiar with parotid surgery in children.

Although our study was limited by its retrospective nature, and the date of initial presentation to the referring physician was often unavailable, there was a general perception that there was often a delay in diagnosis or a failure to recognise the diagnosis at the initial presentation. Nine of our patients had undergone incomplete or inappropriate surgery prior to referral, and this would suggest that a delay in diagnosis had occurred. This finding is in agreement with those of other authors.[2,11,16,17] The rarity of these lesions and their tendency to mimic non-congenital inflammatory conditions is likely to contribute to this diagnostic delay.

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It is generally accepted that the optimal treatment for these lesions is surgical, with complete excision of the tract. This is technically easier in the absence of acute infection, and definitive surgery should therefore be delayed in these cases. In 15 of our cases (83.3 per cent), dissection of the facial nerve was necessary to ensure its protection. This emphasises the vital importance of a correct pre-operative diagnosis, as inappropriate surgery may risk damage to the facial nerve, while incomplete surgery may render further definitive surgery technically more challenging. Of the 15 patients in whom the tract was attached to the cartilaginous external ear, a visible lesion in or near the external auditory meatus was present in 12. In 11 of these patients, there had been evidence of infection or inflammation at this site.

The histological information available from our patients would support the premise that these abnormalities, at least in part, are derived from ectoderm in that they all contained stratified squamous epithelium. The presence of cartilage in all the histological specimens from our patients (including those which would be clinically described as Work type one) would indicate that the histological distinction between the two Work types may not be as clear-cut as previously thought. It is the authors' observation that second arch fistulae tend to secrete mucus, being lined with mucosa. This feature is absent in first arch abnormalities. This may prove a useful clinical sign to distinguish the lesions pre-operatively, since second arch fistulae do not require identification of the facial nerve.

Despite attempts to classify these lesions anatomically and histologically, we have found such classifications of limited clinical use, as when the diagnosis is suspected the tract is exposed surgically and excised completely, with exposure of the facial nerve when appropriate. According to the Work classification, nine of our patients could be classified as type one and four patients as type two. Pre-operatively, all patients were warned of the potential need to expose the facial nerve and of the attendant risks.

- **First branchial cleft anomalies are rare**
- **They are closely related to the facial nerve and external auditory meatus**
- **Examination of the ear is imperative in patients with infection in the peri-auricular area and neck**
- **The surgeon must be prepared to expose the facial nerve when operating on these cases**

Conclusions

Abnormalities of the first branchial cleft are rare and are not confined to the paediatric population. In cases in which the tract communicates with the external auditory meatus, there is often a clinically apparent abnormality in the ear. Clinicians should consider the diagnosis when presented with infected lesions in the peri-auricular and parotid regions as well as the anterior neck, and examination of the ear should be mandatory. Cure requires complete surgical resection of the tract. There is a variable relationship between the tract and the facial nerve. Surgeons treating these abnormalities should be prepared to expose the facial nerve and should obtain appropriately informed consent.

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