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Postać naduszna anomalii pierwszej kieszonki skrzelowej

Supra-auricular presentation of a first branchial cleft anomaly

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Streszczenie

Anomalie kieszonek skrzelowych to zmiany wrodzone, które powstają w wyniku zaburzeń rozwoju aparatu skrzelowego w okresie płodowym. Ze względu na złożoną postać często są błędnie diagnozowane i stanowią wyzwanie w leczeniu. W pracy przedstawiono przypadek 14-miesięcznej dziewczynki z zespołem Goldenhara z prawostronną mikrocją, u której stwierdzono rzadkie, umiejscowione w okolicy nadusznej ujście przetoki prowadzącej do przewodu słuchowego zewnętrznego i komunikującej się z szyją.

Słowa kluczowe: przetoka pierwszej kieszonki skrzelowej, torbiel, zespół Goldenhara, nawracające zakażenie, ropień

Abstract

Branchial anomalies are congenital lesions that occur as a result of incomplete obliteration of the branchial apparatus during foetal development. Due to their complex presentations, they are commonly misdiagnosed and pose a challenge in treatment. This report features a girl, aged 1 year and 2 months, with Goldenhar syndrome with right microtia, presenting with a rare supra-auricular fistulous opening to the external auditory canal, with a communicating tract in the neck.

Keywords: first branchial cleft fistula, cyst, Goldenhar syndrome, recurrent infection, abscess

INTRODUCTION

Anomalies of the branchial arch account for 17% of cervical masses and represent the second most common congenital lesion in the head and neck[1]. First branchial cleft anomalies are relatively uncommon and account for only 1–8% of these defects[2]. The condition has a higher female predilection (69%) compared to the male counterpart (31%)[3]. Von Baer first described the branchial apparatus in 1827. It was further classified histologically into type I and type II by Work[4]. In view of the multifaceted presentations of these anomalies, misdiagnosis leading to a history of repeated incision and drainage procedures is common. First branchial cleft anomalies may masquerade as different morphologies such as a sinus, a fistula or as a cyst occurring in the head and neck. Patients with branchial cleft fistula are typically infants or young children compared to those that present with cysts, who are usually young adults[4].

CASE REPORT

A girl, aged 1 year and 2 months, was referred with recurrent neck abscess and a discharging fistula at the right angle of the mandible since 7 months of age, despite being treated with multiple courses of antibiotics and repeated aspiration. Further history revealed the presence of a fistula below the angle of the mandible since birth. The clinical features were indicative of Goldenhar syndrome, including the presence of right hemifacial microsomia and facial nerve palsy, evidenced by an isolated right depressor anguli oris palsy (Fig. 1 A).

Examination of the neck showed the presence of an inflamed swelling measuring 4 × 2 cm, with an infected discharging fistula below the angle of the mandible. The overlying skin was unhealthy, inflamed and scarred, in keeping with a history of repeated infections (Fig. 1 B).

Computed tomography revealed a fistulous tract communicating between the posterior superior aspect of the right pinna tracking inferiorly just below the right angle of the mandible. There was a heterogeneous collection in the neck within the subcutaneous plane located anterior to the right sternocleidomastoid. The collection measured 1.5 × 1.3 × 2.2 cm (Fig. 2 A, B).

Hearing assessment showed moderate conductive hearing loss on the right ear, with normal hearing on the opposite ear.

Surgical exploration was planned, and the patient underwent excision of the sinus under continuous facial nerve monitoring. A post-auricular incision was made, with inferior extension towards the mandible. To aid the dissection, methylene blue was injected into the fistulous opening in the neck (Fig. 3 A). A fistulous tract was seen running parallel along the external auditory canal to the neck, anterior to the sternocleidomastoid, above the level of the hyoid (Fig. 3 B). The tract measured 9 cm and was excised as a whole (Fig. 3 C).

Histopathological examination described a tract lined by keratinised stratified squamous epithelium with focal adnexal structures. Focal ulcerative areas were seen along the tract, with moderate to dense lymphoplasma and neutrophilic aggregates. The findings confirmed the diagnosis of a first branchial cleft fistula (Fig. 4 A, B).

No evidence of relapse was observed over a 4-month postoperative period.

DISCUSSION

Between weeks 4 and 5 of gestation, the branchial apparatus is formed. It develops four ectodermal grooves called clefts, and five endodermal pouches. The endoderm in turn invaginates and migrates towards the ectoderm, forming branchial arches. The first branchial cleft is a recess that lies between the first and second branchial arches. The cleft matures and forms the external auditory canal and the lateral surface of the tympanic membrane[5]. Any incomplete closure of the

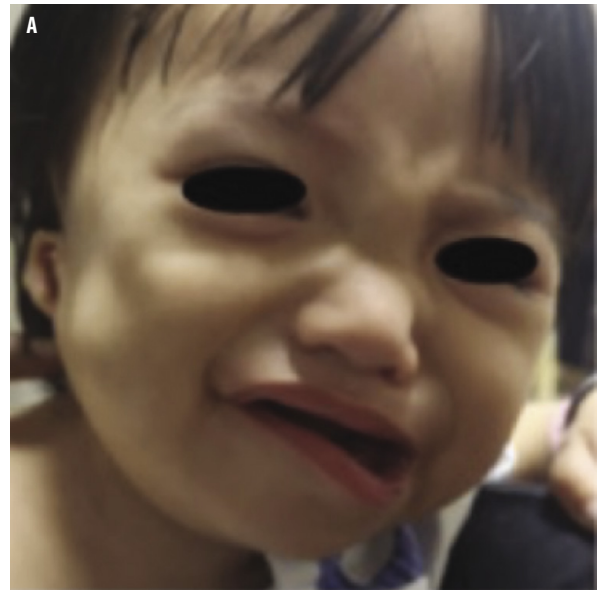

Fig. 1 A. Goldenhar syndrome, with right hemifacial microsomia and facial nerve palsy, evidenced by an isolated right depressor anguli oris palsy

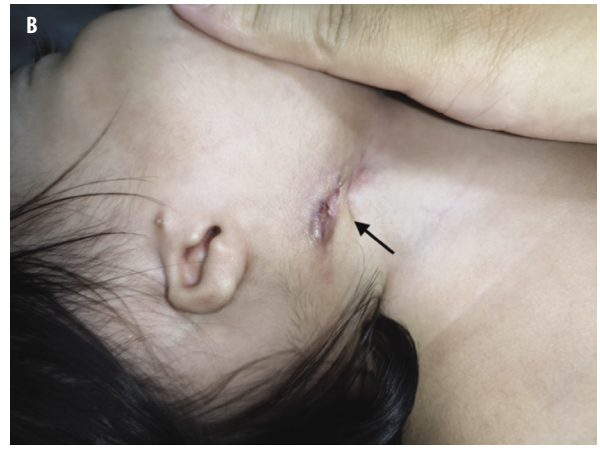

Fig. 1 B. Left swelling measuring 4 × 2 cm, with a discharging fistula at the right angle of the mandible. Notice the overlying skin scarring (black arrow)

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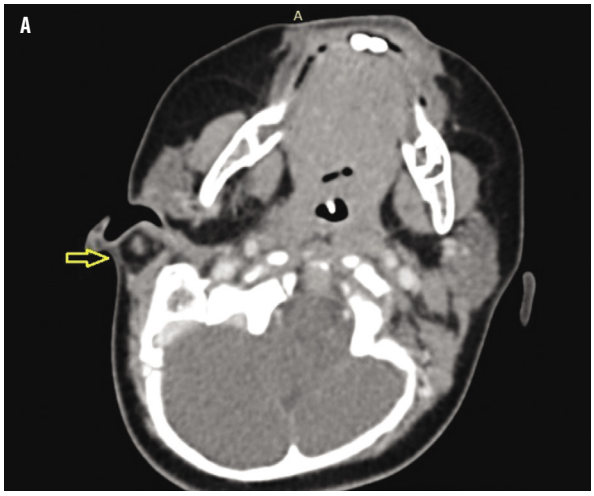

Fig. 2 A. Axial contrast-enhanced computed tomography demonstrating a fistulous tract posterior to the right pinna (yellow arrow)

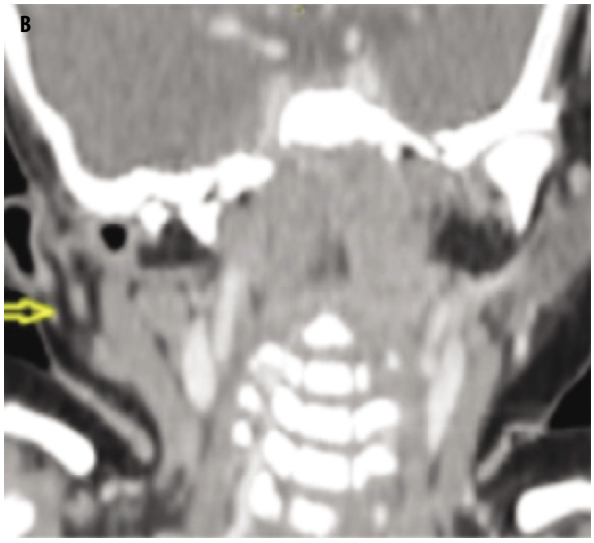

Fig. 2 B. Coronal contrast-enhanced computed tomography demonstrated a fistulous tract extending inferiorly to the right angle of the mandible, with heterogeneous collection in the subcutaneous plane (yellow arrow)

ectodermal part of the first branchial cleft may result in either a sinus, cyst or fistula. Its common external opening either sinus or fistula are the external auditory canal (40%), upper neck anterior to the sternocleidomastoid (32.5%), concha (20%) and post-auricular (7.5%)[2].

They have been categorised into type I and type II lesions based on anatomical and histological features. Arnot[6] classified first branchial cleft anomalies into type I and type II based on their clinical presentation. Type I defects were described as sinuses or cyst openings; medial, posterior to pinna and concha or at the angle of the mandible. The tract may run parallel to the external auditory canal. Patients may present as young adults with recurrent pre-auricular swellings or abscesses.

Type II anomalies are often seen in early childhood, where a fistulous tract may be seen running from the floor of the external auditory canal inferiorly below the mandible to above the level of the hyoid. Patients may have recurrent otorrhoea, thus, a detailed otological examination is warranted. Examination may reveal a pit visible in the external auditory canal at the entrance of the fistula, or there may be a complete absence of signs in the ear canal. Patients

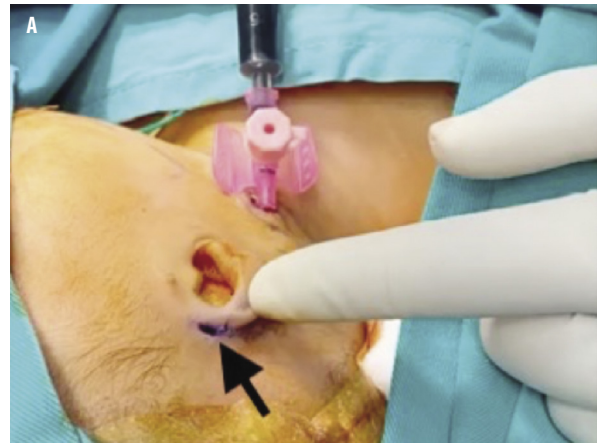

Fig. 3 A. Supra-auricular delineation of the tract with methylene blue dye (black arrow)

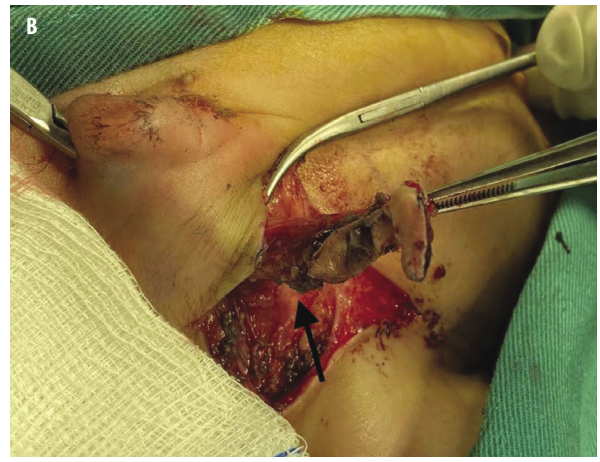

Fig. 3 B. Dissection of the cleft via a low post-auricular incision with extension to the neck (black arrow)

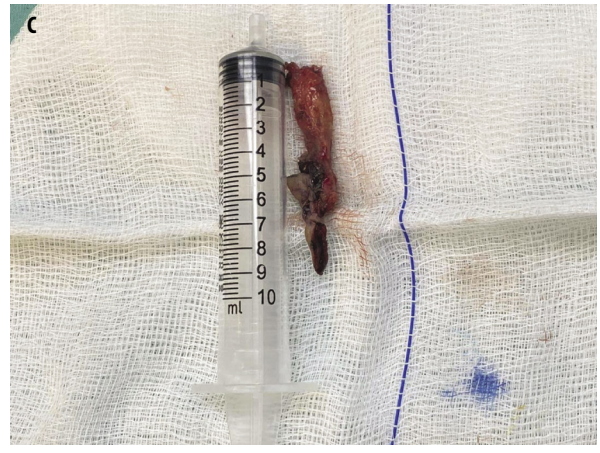

Fig. 3 C. Cleft excised completely, measuring 9 cm in length

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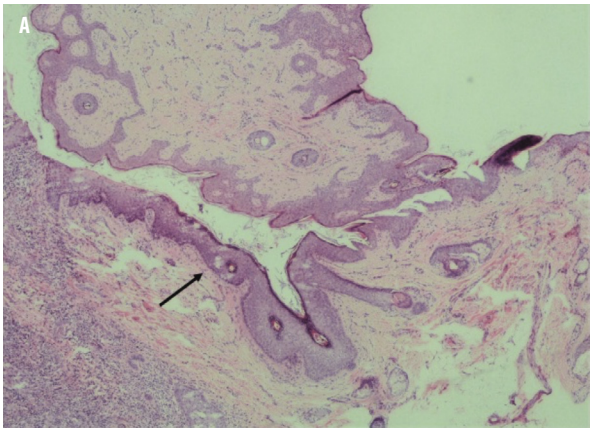

Fig. 4 A. Section showing tissue lined by keratinised stratified squamous epithelium with a tract (black arrow) extending from the surface

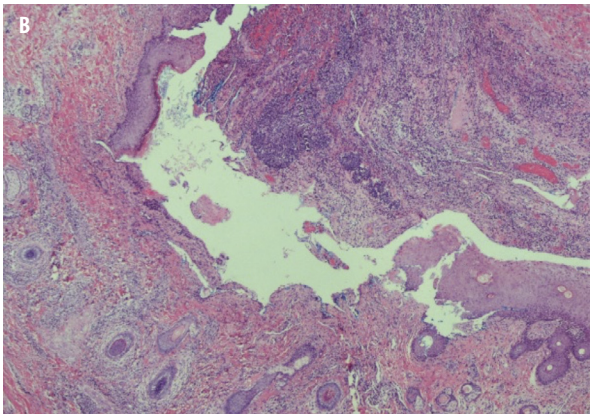

Fig. 4 B. Section showing the cleft tract lined by keratinised squamous epithelium with the presence of adnexal structures such as hair follicles and sebaceous glands

may also present with a recurrent discharging fistula and abscesses just below the mandible.

This mirrors the intra-operative findings in the patient reported above, with a variation in the course of the supra-auricular fistula tract. Type II anomalies are more common than type I. Middle ear structures are normal in most cases. For type II lesions, early identification of the facial nerve trunk during dissection is of paramount importance, as its relationship cannot be predicted precisely pre-operatively. The relation of the fistula and facial nerve varies, as it may lie medial (37%), lateral (41%) or in between the facial nerve branches (22%)[2]. It has been found that the tract tends to be situated deep to the facial nerve in subjects presenting before the age of 6 months old. Work histologically classified first branchial cleft anomalies into type I and type II.

Type I lesions are lined by squamous epithelium, with absent mesenchyma. Type II lesions involve the duplication of membranous and cartilaginous parts of the ear canal. They contain skin and adnexal structures, which was seen in this patient's biopsy report.

The management of a branchial cleft fistula is typically based on surgical excision beyond 3 to 6 months of age. This is due to the patients' underdeveloped auricular cartilage and mastoid. In addition, this allows the resolution of an ongoing infection that is commonly seen. Broad spectrum systemic antibiotics and aspiration are preferred over incision and drainage so as to avoid further distortion to the surgical plane[7].

The impression of an abscess over the fistulous tract may lead to inadvertent incision and drainage procedures. This misdiagnosis often leads to scarring, which makes excision of the tract challenging, keeping in mind the variation in its course and close proximity to the facial nerve, which lies more superficially in paediatric cases. If the diagnosis is unclear, biopsy of the cyst wall is done to aid in differentiating between an infected branchial cleft cyst and simple bacterial lymphadenitis[8].

Surgery is aimed at complete excision of the lesion with preservation of facial nerve function. Wide exposure of the lesion is pivotal for safe excision, and modifications of surgical incisions may be required, depending on the course of the lesion, as seen in our case. Failure to identify and expose the facial nerve has been shown to be associated with a greater incidence of post-operative facial nerve palsy[2].

CONCLUSIONS

First branchial cleft anomalies are known to have a myriad of presentations, which makes the diagnosis challenging. A high index of suspicion of these lesions should be raised in patients with a history of recurrent otorrhoea, and repeated incision and drainage procedures of pre-auricular, post-auricular or upper neck swellings. This is to prevent delayed diagnosis and suboptimal patient treatment.

Conflict of interest

The authors do not report any financial or personal connections with other persons or organizations which might negatively affect the content of this publication and/or claim authorship rights to this publication.

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