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Case Report

First branchial cleft fistula (Work Type 2) with an internal opening to the Eustachian tube: Case report and review of literature

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ABSTRACT

First branchial cleft anomalies are rare congenital defects of the head and neck. This case report presents a 12-year-old patient with a draining cutaneous pit approximately 1-cm anterior and 5-mm inferior to the right angle of the mandible. Imaging revealed a fistula between the cutaneous pit and Eustachian tube. Further characterization with methylene blue injection into the cutaneous pit resulted in spillage through the right Eustachian tube. Surgical excision of the fistula revealed a cylindrical structure comprised of ectodermal and mesodermal features that most likely represented a Work Type 2 first branchial cleft fistula.

Introduction

Branchial cleft anomalies account for about 30% of all congenital head and neck lesions [1]. Anomalies of the first branchial cleft make up less than 8% of all branchial cleft anomalies and occur at an estimated rate of 1 per million people per year [2]. First branchial cleft anomalies can occur anywhere along the course of the first branchial arch structures [3]. The defect is typically encountered between the bony cartilaginous junction of the external auditory canal and either extends inferiorly to the submandibular triangle or to the preauricular area, depending on the type, classified by Work as Type 1 or 2 [3]. This paper reviews the relevant embryology and the difference between Work Types 1 and 2 first branchial cleft anomalies. Additionally, it will present the differences between a case of a presumed Type 2 first branchial cleft fistula with an opening to the Eustachian tube and 2 other cases with similar findings that have been reported in the literature.

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Case report

A 12-year-old female presented to the Otolaryngology Department at a Tertiary Care Military Medical Center with complaints of a draining cervical pit. Per the child's mother, the pit had been present since birth but had only started to drain within the last year. There had never been any associated erythema, pain, purulent drainage, or other signs of infection. The patient described the drainage as spontaneous, random, and mucoid in nature. She found this socially disturbing and desired surgical excision.

On physical examination, the child was a healthy-appearing preadolescent female in no distress. She was able to point out a small cutaneous dimple located approximately 10-mm anterior and 5-mm inferior to the angle of the right mandible. There was no drainage and no associated mass lesion. The remainder of the head and neck exam was normal.

Given the location of the lesion and its proximity to the marginal mandibular nerve, further workup was necessary. After discussion with a pediatric radiologist, a contrast study was performed to outline the extent of the fistula. Placing a lubricated 23G intravenous catheter into the dimple, low osmolality nonionic iodinated contrast was injected under fluoroscopy revealing a fistulous tract extending into nasopharynx, which induced coughing during the injection (Fig. 1). The patient was immediately transferred to obtain a 3D-CT fistulogram to delineate the course of the tract. CT demonstrated an opacified tract from the skin surface at the anterior inferior aspect of the right mandibular angle traversing the deep soft tissues posterior to the parotid mandible and through the parapharyngeal space into the mucosal pharyngeal space extending toward the torus tubarius of the Eustachian tube (Fig. 2).

The patient and her parents were counseled that surgical excision would pose a significant risk to the marginal mandibular nerve. However, they elected for excision. Intraoperatively, methylene blue dye mixed with fibrin glue was injected into the cervical opening of the fistula. Nasopharyngoscopy was performed and revealed the dyed solution extruding from the right Eustachian tube orifice during injection (Fig. 3). An incision was made in a skin crease approximately 2 finger breadths below the border of the mandible. Nerve monitoring was performed throughout the case. Subplatysmal flaps were raised and the inferior border of the submandibular gland was identified. The submandibular gland was displaced anteriorly and we were able to palpate the tract deep to the body of the mandible. A small elliptical incision was then made around the cutaneous pit and the tract was followed until it could be delivered through the primary incision. The mid portion of the tract was noted to be cartilaginous in nature. The marginal mandibular branch of the facial nerve was adherent to the fistula tract and was dissected free and preserved. Blunt dissection was used to follow the fistula tract into the parapharyngeal space to the skull base (Fig. 4). At the skull base, a right angle clamp was placed on the tract, the tract was transected (Fig. 5) and the cranial portion was tied off with silk suture. A suction drain was placed for 24 hours postoperatively, and the patient was discharged home the following day. She initially had paralysis of the marginal mandibular nerve, but over the first postoperative month, nerve function recovered. She had no Eustachian tube dysfunction postoperatively.

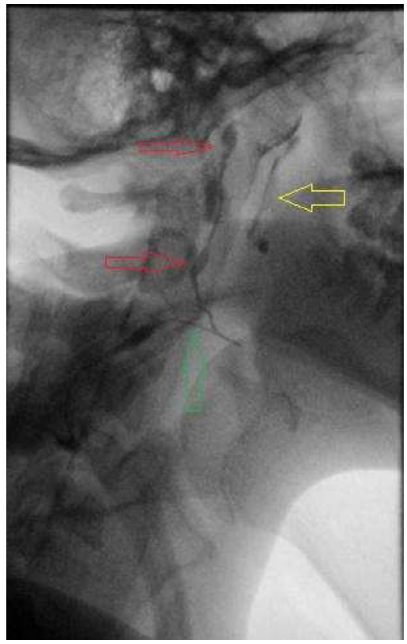

Fig. 1 – Spot fluoroscopic image demonstrates the 23G angiocatheter (*green arrow*) inserted into the submandibular angle dimple. Low osmolar nonionic iodinated contrast was gently injected through the catheter, demonstrating a 2 x 2 mm opacified tract (*red arrows*) with variable width extending cephalad draining into the retropharynx (*yellow arrow*) at the level of the adenoids. Patient was sent to CT for definitive pathway identification. (Color version of figure is available online.)

Our patient most likely presented with a Work Type 2 first branchial cleft fistula based on the location of the fistula openings and histology of the specimen. The opening of the fistula at the submandibular triangle is characteristic of a Type 2 first branchial cleft anomaly [3,4]. There are 2 reported cases of Type 2 first branchial cleft fistulas that open to the Eustachian tube found after a thorough review of the literature. Both cases were reported by Liu et al [5].

Discussion

Embryology

The development of the head and neck is important to understand in identifying congenital anomalies as the structure and orientation of the branchial arches and branchial pouches will help providers differentiate the origin and location of such defects. Weeks 4-5 of gestation mark the formation of the branchial arches and clefts that are the embryologic precursors to many structures of the head and

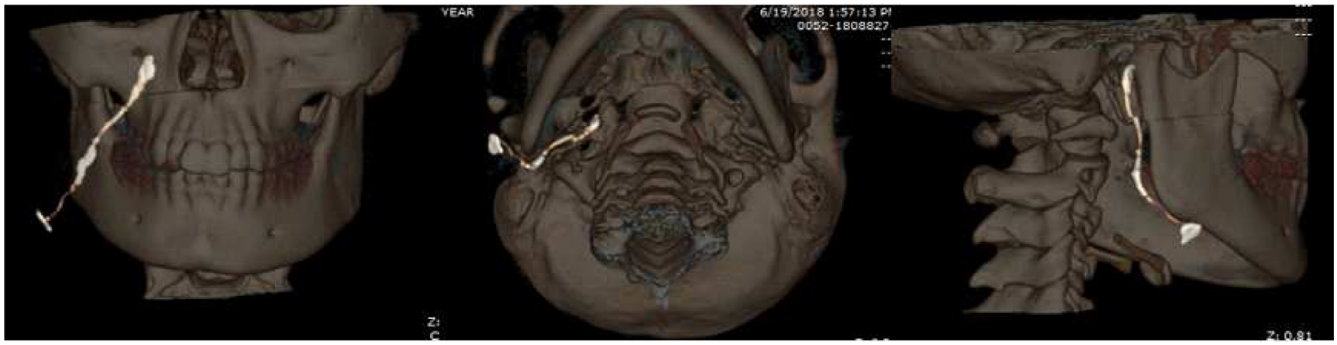

Fig. 2 – Coronal, caudocranial, and right oblique 3D soft tissue subtraction-reconstructed images demonstrate the fistula extending from the skin surface to the deep soft tissues anterior to the parotid space, posterior to the masticator space through the parapharyngeal space into the mucosal pharyngeal space extending toward the torus tubarius of the Eustachian tube.

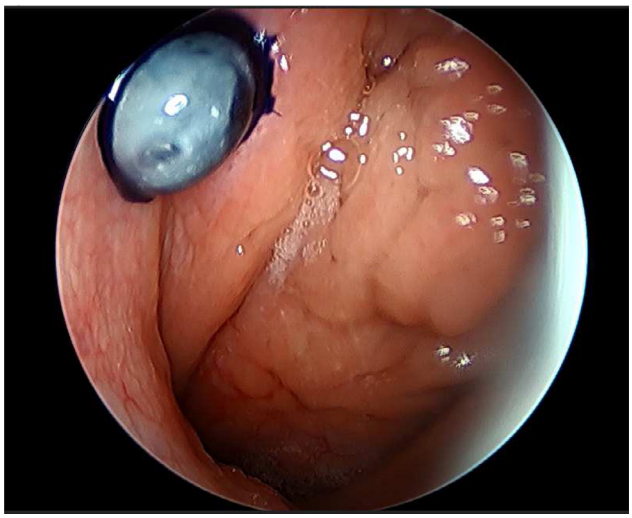

Fig. 3 – Injection of methylene blue dye into the cutaneous pit during nasopharyngoscopy verified communication of the defect with the Eustachian tube.

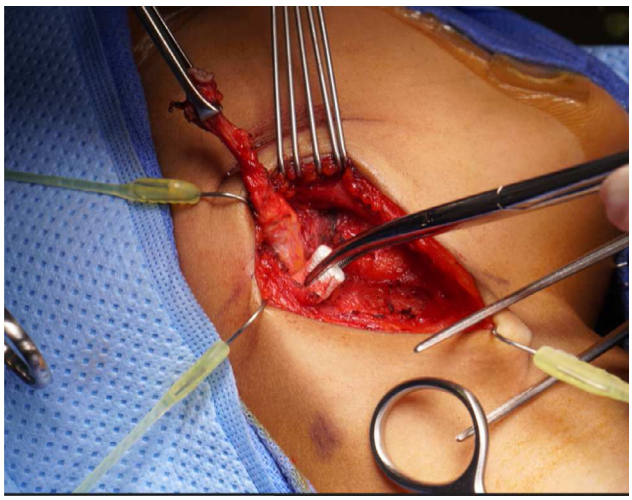

Fig. 4 – Intraoperative photo of fistula excision.

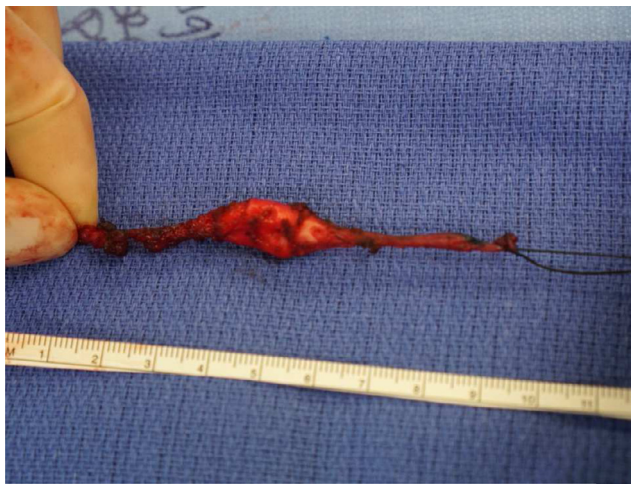

Fig. 5 – The complete fistula measured approximately 9 cm in length when taught and was noted to have a cartilaginous midsection.

neck [4]. During this period, 4 well-defined masses begin to develop along with 2 rudimentary masses, bilaterally, in a craniocaudal progression along the branchial foregut [3,4,6]. Endoderm lines the internal/pharyngeal aspect, while ectoderm lines the branchial arches and clefts, or external aspect [3,4]. Mesenchyme consisting of mesoderm and neural crest cells fills the space between the endoderm and ectoderm [3,4]. The outer portions of these masses are termed branchial arches, which contain ectoderm and mesoderm. In total, there are 6 pairs of branchial arches from the 6 masses that develop, with the fifth arch being a rudimentary arch that never fully develops [4]. These are separated by 4 indentations called branchial clefts [3,4]. The inner outpouchings of the masses that contain the endodermal epithelium are called branchial pouches [4]. The branchial clefts and branchial pouches undergo significant remodeling with most of the embryologic tissue being obliterated by the mesenchyme during

remodeling to allow for the development of various head and neck structures [3].

The mesodermal portion of the branchial arches will ultimately develop into the muscle, bone, cartilage, and connective tissue of the head and neck [4]. Each branchial arch also has its own associated artery and cranial nerve [3,4]. The branchial pouch endoderm will develop many structures within the pharynx including the Eustachian tube, lining of the tonsillar fossa and parathyroid glands [4]. The endoderm that lines the branchial pouches at the apex invaginates toward the ectodermal lining of the branchial clefts [4]. This contact is an important aspect of remodeling due to the proximity of the 2 and the possibility that improper remodeling, or incomplete obliteration, may take place. The only branchial cleft that persists in normal development is the first branchial cleft [7]. In normal development, the remaining branchial clefts are obliterated by the growth and differentiation of the surrounding branchial arches and do not develop into any adult structures [4]. The first branchial cleft is responsible for the external layer of the tympanic membrane, the external auditory canal, and the conchal cartilage [8]. The first branchial pouch will develop into the tubotympanic recess, which includes the inner layer of the tympanic membrane, the tympanic cavity, and the Eustachian tube [3]. During development, the first branchial pouch will fuse with the first branchial cleft to form the complete tympanic membrane with mesoderm cells lining the middle portion creating the fibrous layer of the tympanic membrane [7]. The first branchial arch notably develops into the mandible, maxilla, malleus, incus, sphenomandibular ligament, maxillary artery, trigeminal nerve, and the muscles of mastication [3,8]. The remodeling that takes place is not completely understood, but has been known to create the spectrum of branchial cleft anomalies [4]. Branchial anomalies are generally identified by the associated embryologic arch or pouch precursor of the local anatomy [6]. These anomalies are thought to persist as a result of incomplete obliteration of the clefts due to incomplete fusion of the ventral portion of the first and second branchial cleft arches [8].

Classification of first branchial anomalies

The identification of the embryologic structure involved is typically determined by anatomy [6]. The incomplete fusion of the first and second branchial arches that results in branchial cleft anomalies occurs during the development of the parotid gland and facial nerve [8]. This results in the fistula location being periauricular or near the angle of the mandible [8].

Anomalies of the first branchial arch are typically associated with the external auditory canal and middle ear space [3,9]. The current means of differentiation between types of first branchial cleft defects have not significantly changed since they were classified by Work and further described by Olsen et al [9–11]. Current classification lists the first branchial anomalies as Work Type 1 or 2.

Work Type 1 first branchial cleft anomalies present as cystic periauricular masses. They are the result of a duplication of the membranous external auditory canal and are comprised only of ectodermal tissue [3]. Histologically, these cysts consist of squamous epithelium. The cyst will classically run a course along the external auditory canal [3].

When identifying a Work Type 2 first branchial cleft anomaly, it is important to classify it as a branchial cleft cyst, sinus or fistula as described by Olsen et al [9]. The primary means of differentiating these is the number of openings to either the lumen of the foregut or the skin [11]. A cyst lacks any openings that may drain a fluid collection [11,12]. A fistula and a sinus both form tube-like structures. The primary difference is that a sinus has a single opening into a cavity or surface, whereas a fistula has two openings between epithelialized surfaces with 1 opening typically being at the skin [4,12]. These first branchial cleft defects consist of both ectodermal and mesodermal tissues, which include squamous epithelium, cartilage, and adnexal structures [3]. These anomalies are classically identified as a submandibular mass or cutaneous pit in the submandibular triangle near the angle of the mandible [3,4].

Management pearls

Surgical excision of a branchial cleft defect is the only definitive treatment [6]. A 35-year retrospective study by Al-Mufarrej et al reported that 3.6% of branchial arch anomalies in 358 patients recurred within a mean of 4 years [13]. The branchial arch defect type, the use of imaging, and the extent of excision did not affect the recurrence rate. The only finding that appeared to affect the rate of recurrence was previous incision and drainage, which increased recurrence 3.6-fold [13]. In general, about 20% of patients will undergo surgery after they have had a previous infection [6]. This is an important consideration in the management of acute infections. Primary treatment should involve antibiotics with a consideration of fine needle aspiration for cystic loculations. An incision and drainage should be considered in cases that cannot be managed with antibiotics or fine needle aspiration alone [6]. Surgical excision should be considered after the infection has resolved [6]. A literature review by D'Souza et al reports that temporary facial nerve palsy occurred in about 20% of cases, while permanent facial nerve palsy occurred in about 3% of cases [14]. Due to the potential morbidity, surgical resection of first branchial arch anomalies should include careful dissection to preserve the facial nerve. Additionally, surgery should include the excision of any involved skin or cartilage of the middle ear to prevent recurrence [5,6].

Explanation of patient findings

The first case from the literature involved a fistula that opened anteriorly into the Eustachian tube and traveled intimately with the external auditory canal. It traversed through the cartilage of the canal and under the skin while traveling superficially to the trunk of the facial nerve [5]. This path along the external auditory canal is a classically described pathway of a typical Work Type 1 first branchial cleft fistula [10]. The diagnosis of a Work Type 2 fistula was made on the basis of the tissue present within the tract. The deep portion of the fistula required dissection off of the deep lobe of the parotid gland [5]. Our patient had a significantly different trajectory of the fistula tract. The tract was posterior to the parotid and masticator spaces, which allowed for the cutaneous pit to open to the anteroinferior border of the mandible and the fistula

to pass deep to the facial nerve. The second case involved a 14-month-old patient with a diagnosis of atypical hemifacial microsomia with first and second branchial arch syndromes [5]. This child had multiple developmental abnormalities including an absent facial nerve on the affected side and a stenotic left external auditory canal [5]. The branchial cleft fistula in this patient followed a similar path to the other patient described in the same paper. The fistula passed through the cartilage of the external auditory canal and then under the skin before appearing as a 1-mm cutaneous opening near the lobule posterior to the mandibular ramus [5]. Again, the primary difference between the fistula of this patient and the patient presented in this case report is the path of the fistula. Our patient was nonsyndromic and had a fistula that was not coursing along the external auditory canal. The difference in fistula path is likely the result of differences in embryologic development. There are several theories that explain the development of branchial cleft anomalies which include the branchial apparatus theory, cervical sinus theory, thymopharyngeal theory, and inclusion theory [10]. The most widely accepted theory for branchial cleft defects involves incomplete obliteration of the second, third, and fourth clefts and pouches [10]. This incomplete obliteration allows for the embryologic tissues to persist creating a branchial cleft anomaly [15]. The outlier would be the Type 1 branchial cleft anomaly that is due to the duplication of the external auditory canal [3].

While this anomaly is a rare event with incomplete obliteration as the cause of our patient's Type 2 first branchial fistula, the relationship between the first branchial cleft and branchial pouch allows for the most logical explanation of how this patient's fistula developed. During normal embryologic development, the first branchial cleft and first branchial pouch fuse to form the tympanic membrane while the Eustachian tube develops from the endoderm of the first branchial pouch [4,7]. In this patient, a second abnormal fusion event likely occurred as a result of abnormal mesenchymal involution between a portion of the first branchial pouch and first branchial cleft [5]. This led to an incomplete obliteration by the mesenchyme and possible persistence of other mesodermal and ectodermal cells within the area. This second fusion site was likely caudal to the apex where the tympanic membrane is formed by the fusion of the first branchial cleft and pouch. This allowed for mesoderm trapping and eventual development of a cartilage structure in a location that it normally does not belong. If this fusion event was caudal to the apices of the first branchial cleft and pouch at the tympanic membrane fusion point, it could explain why the fistula entry point was inferior to the tympanic cavity and opened into the Eustachian tube. This likely created a very similar process as if it were developing into another external auditory canal, which is the reason it persisted and did not obliterate. This theoretical duplication of the external auditory canal would normally suggest a Type 1 first branchial cleft cyst. However, the types of tissue within the fistula tract included a cartilaginous tube structure and squamous epithelium suggesting the fistula had mesodermal and ectodermal derived tissues. The presence of multiple tissues suggests that this is a Type 2 first branchial cleft fistula [3,4].

First branchial cleft anomalies are rare congenital defects. Proper identification of these defects with imaging will allow practitioners to develop management strategies that can reduce the likelihood of recurrence by limiting incision and drainage to cases of acute infection that require more than antibiotic therapy and improve intraoperative safety by limiting the number of operations and reducing the risk of nerve injury. In instances of branchial cleft fistulas with unusual presentations, as with this case, the use of methylene blue dye injection into the defect may help verify the suspected path of a fistula or sinus [5,16]. Surgery remains the only definitive treatment for a branchial cleft anomaly.

Supplementary materials

Supplementary material associated with this article can be found, in the online version, at doi:10.1016/j.radcr.2019.03.036.

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