International Journal of Science and Research Archive
eISSN: 2582-8185
Cross Ref DOI: 10.30574/ijsra
Journal homepage: https://ijsra.net/

(CASE REPORT)

A comprehensive approach to the diagnosis and surgical management of first branchial cleft anomaly: A case report and literature review

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International Journal of Science and Research Archive, 2026, 19(02), 1508-1513

Publication history: Received on 23March 2026; revised on 26 May 2026; accepted on 28 May 2026

Article DOI: https://doi.org/10.30574/ijsra.2026.19.2.1197

Abstract

Background: First branchial cleft anomalies (FBCAs) are rare congenital malformations that result from incomplete obliteration of the first branchial cleft during embryogenesis. They often present with recurrent infections, swelling, and purulent drainage in the periauricular or parotid regions, leading to challenges in diagnosis and management.

Case Presentation: An 11-year-old girl with no significant personal or family medical history presented with recurrent right parotid region inflammation and intermittent purulent drainage. Initial management at an outside facility included incision and drainage, but the condition progressed to a persistent cutaneous fistula. Imaging studies, including high-resolution cervical ultrasonography and contrast-enhanced MRI, revealed a tubular fistulous tract extending from the right parotid region toward the skin opening, with surrounding chronic inflammation and a cystic formation along the tract. Surgical exploration confirmed the tract's deep relationship with the facial nerve. The patient underwent a superficial parotidectomy, with successful facial nerve preservation and no recurrence at 12 months of follow-up.

Conclusion: FBCAs, though rare, require early diagnosis and timely surgical intervention to prevent recurrent infections and nerve damage. Advanced imaging modalities such as MRI are essential for accurate preoperative assessment and surgical planning. Complete excision with facial nerve preservation is the gold standard for treatment, offering excellent long-term outcomes when managed in specialized centers.

Keywords: First Branchial Cleft Anomaly; Pediatric; Facial Nerve Preservation; Parotid Gland; Fistula; MRI; Surgery

1. Introduction

First branchial cleft anomalies (FBCAs) are rare congenital malformations that arise from incomplete obliteration of the first branchial cleft during embryonic development. These anomalies account for less than 8% of all branchial cleft defects, making them a relatively uncommon clinical entity. FBCAs typically present in early childhood and are most often diagnosed between the ages of 2 and 5 years, though they can be detected at any age. The condition can manifest in various forms, including cysts, sinuses, or fistulas, depending on the extent of cleft failure and the degree of tissue differentiation involved in the malformation. Although the external manifestations of FBCAs are usually located in the periauricular region or near the parotid gland, their variable presentation often leads to misdiagnosis and delayed treatment, particularly when there is recurrent inflammation or infection [1], [2].

The first branchial cleft develops during early embryogenesis and is responsible for forming the external auditory canal (EAC) and parts of the auricle. When anomalies occur, they are typically seen as abnormal tracts or cysts that can extend

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toward the skin surface, causing recurrent infections or discharge. These anomalies are often mistaken for other conditions, such as recurrent otitis media, cystic parotid tumors, or chronic abscesses, which complicates early diagnosis and proper treatment. Advanced imaging, such as high-resolution ultrasound and contrast-enhanced MRI, is crucial for accurate assessment and surgical planning, as it helps in visualizing the tract and understanding its relationship to nearby structures like the facial nerve and parotid gland [3], [4].

Surgical intervention remains the cornerstone of treatment, with complete excision being necessary to prevent recurrence and minimize complications. However, the proximity of the facial nerve to the branchial cleft anomaly poses a challenge during surgery. Mismanagement during dissection can lead to facial nerve injury, resulting in temporary or permanent facial paralysis. Therefore, a thorough understanding of the anatomy and preoperative planning using modern imaging modalities is essential for successful outcomes. Previous studies have emphasized that early diagnosis and surgical intervention in non-inflammatory stages can significantly reduce the risk of complications and improve the prognosis for affected patients [5], [6].

This case report details the presentation, diagnostic workup, surgical management, and postoperative recovery of an 11-year-old girl with a first branchial cleft anomaly. The goal is to emphasize the importance of early diagnosis, appropriate imaging, and expert surgical management in achieving favorable outcomes in pediatric patients with this rare condition.

2. Case presentation

An 11-year-old girl with no relevant personal or family medical history was referred to the Otorhinolaryngology departement for recurrent inflammatory episodes involving the right parotid region, complicated by intermittent purulent drainage. The patient's first episode occurred in early childhood, manifesting as a deep soft-tissue infection in the right parotid area, without otalgia, otorrhea, hearing complaints, or other otologic symptoms. Initial management at an outside facility consisted of incision and drainage combined with systemic antibiotics. Despite repeated treatments, the clinical evolution was unfavorable, progressing to a persistent cutaneous fistula with episodic purulent discharge and progressive scarring.

At presentation, inspection revealed a retracted, cosmetically deforming scar in the right parotid–mandibular angle region, centered on a small cutaneous orifice actively draining purulent material (Figure 1). Palpation demonstrated induration along a short subcutaneous tract without a clearly palpable encapsulated mass. Otoscopy showed a normal external auditory canal and a normal tympanic membrane, with no visible pit or discharge within the canal. No cranial nerve deficit was present preoperatively.

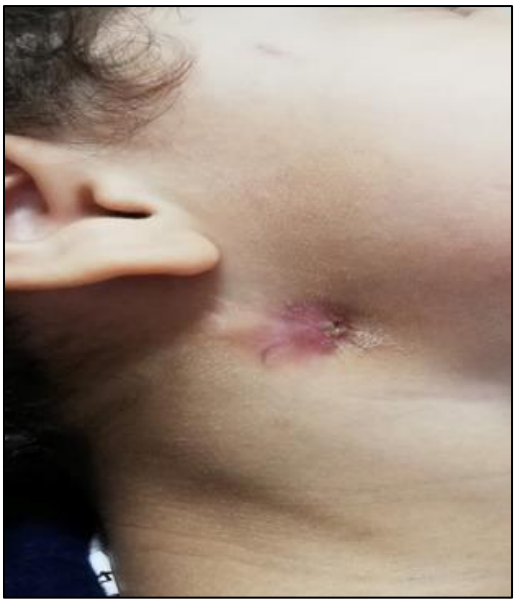

Figure 1 Clinical presentation of the patient showing a retracted, cosmetically deforming scar in the right parotid–mandibular angle region, centered on a small cutaneous orifice actively draining purulent material. This image illustrates the characteristic external manifestation of a first branchial cleft anomaly with episodic purulent discharge

High-resolution cervical ultrasonography, performed as the first-line imaging modality, revealed a linear to tubular hypoechoic structure within the superficial lobe of the right parotid region, extending toward the cutaneous opening. This sonographic appearance, consistent with first branchial cleft anomalies, showed duct-like or strip-like hypoechoic structures in the parotid. Surrounding the tract was mild hyperechogenicity of adjacent fat planes, indicative of chronic inflammation. No intraparotid sialolithiasis, ductal dilatation of Stensen's duct, or suspicious solid nodules were observed.

A contrast-enhanced MRI of the parotid and upper neck was performed to map the lesion before definitive surgery. The MRI revealed a tubular fistulous tract extending through the right parotid tail toward the skin opening, surrounded by inflammatory changes, with no signs suggestive of neoplasia. On T1-weighted images, the tract appeared hypointense, while on T2-weighted images, it showed hyperintensity, consistent with fluid-filled structures. Additionally, a cyst was identified along the malformation's trajectory, further supporting the diagnosis of a first branchial cleft anomaly (Figure 2).

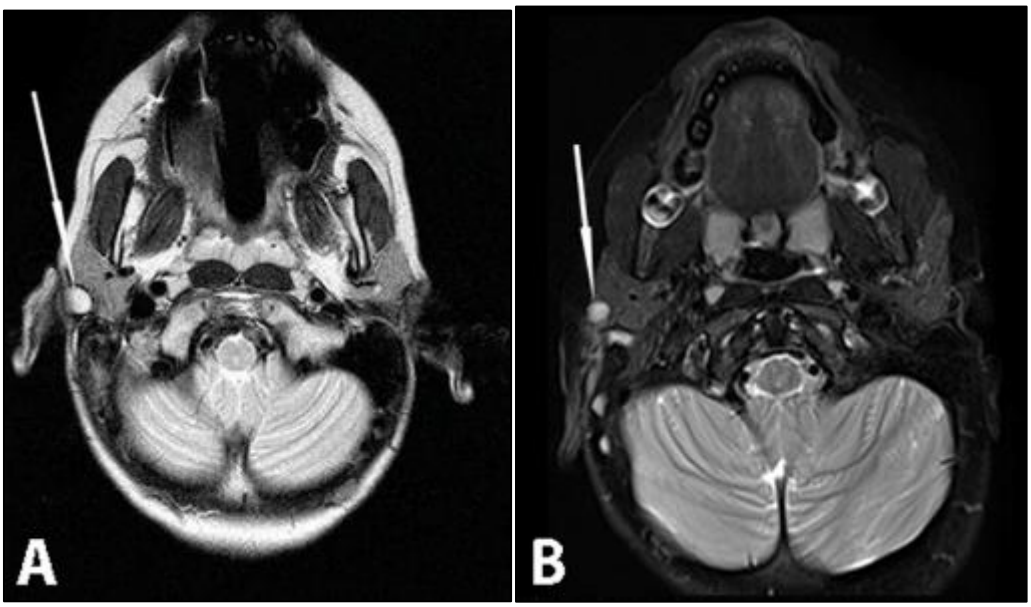

Figure 2 MRI findings of the fistulous tract in the right parotid region. A: T1-weighted image showing a hypointense tubular tract extending toward the skin opening, with a cystic formation indicated by the arrow. B: T2-weighted image revealing the same tract with hyperintensity, consistent with fluid-filled structures, and the cystic formation again indicated by the arrow

Definitive surgery was performed under general anesthesia. A superficial parotidectomy via an anterograde approach was undertaken. The facial nerve trunk was identified, dissected, and preserved. The fistulous tract was meticulously followed; it passed deep to the facial nerve and traversed parotid tissue, terminating beneath the posterior belly of the digastric muscle, where it was ligated and excised en bloc (Figure 3). Postoperative recovery was uneventful, with no facial weakness, salivary fistula, or wound complication. At 12-month follow-up, clinical examination showed satisfactory healing and no evidence of recurrence.

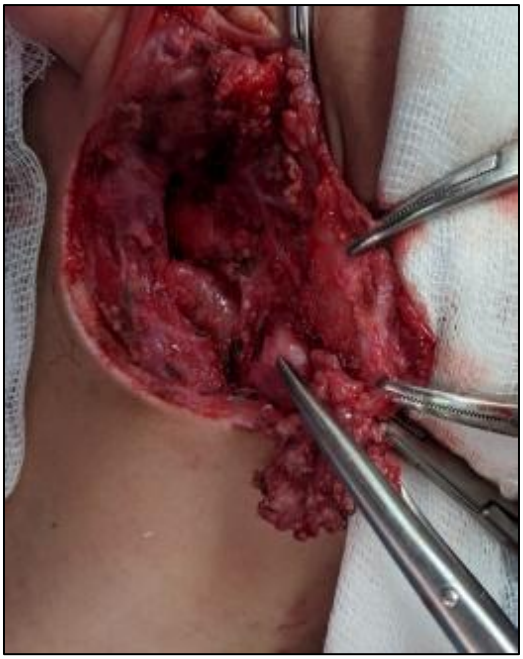

Figure 3 Intraoperative view showing the dissection of the fistulous tract within the right parotid region. The tract is carefully traced toward its termination beneath the posterior belly of the digastric muscle, with the facial nerve clearly identified and preserved during the procedure

The final histology revealed a tract lined with keratinized stratified squamous epithelium, consistent with the nature of first branchial cleft anomalies. The tract also showed associated chronic inflammation, confirming the presence of a long-standing inflammatory process. cartilage was noted within the tract walls, as well as skin adnexal structures.

3. Discussion

First branchial cleft anomalies (FBCAs) are rare congenital malformations that occur due to incomplete obliteration of the first branchial cleft during embryonic development. These anomalies typically present with a range of clinical features, including recurrent swelling, purulent drainage, or chronic infections in the periauricular and parotid regions. FBCAs can present as sinuses, cysts, or fistulas, and their complexity often leads to diagnostic challenges and delayed treatment. The clinical presentation and management of FBCAs vary significantly across different studies, and our case highlights both the common features and the challenges of diagnosing and treating these anomalies in pediatric patients.

The clinical presentation of our patient, an 11-year-old girl with recurrent right parotid region inflammation and persistent purulent drainage, aligns with the typical findings reported in recent studies. The patient's presentation, complicated by previous untreated infections and a persistent cutaneous fistula, mirrors the situation reported in previous studies, which emphasized the challenges of diagnosing FBCAs due to their nonspecific symptoms and frequent misdiagnosis. Our findings are consistent with those from studies that have noted the prevalence of recurrent infections leading to suboptimal initial management, often involving incision and drainage procedures [1], [2].

Imaging plays a vital role in diagnosing and managing FBCAs. In our case, high-resolution cervical ultrasonography was used as the first-line imaging modality, revealing a linear hypoechoic structure that aligned with the typical sonographic appearance of first branchial cleft anomalies. The imaging characteristics were similar to those described in other studies, where ultrasound was employed to identify the tract's course and surrounding inflammatory changes in pediatric cases [5]. However, ultrasonography alone does not always provide a comprehensive assessment, especially in complex cases where the tract may extend deep into surrounding tissues. This limitation was highlighted by previous research, which demonstrated that cross-sectional imaging, particularly MRI, is more effective in delineating the full extent of the lesion and its anatomical relationships, especially when it involves critical structures like the facial nerve [2], [6].

The use of contrast-enhanced MRI was essential in our patient's case. MRI allowed us to map the lesion's full trajectory, showing a tubular fistulous tract with characteristic hyperintensity on T2-weighted images, consistent with fluid-filled structures, and a cystic component along the tract's path. This finding is consistent with the imaging characteristics

observed in similar cases, where MRI was used to assess the tract's relationship to the facial nerve and surrounding tissues [8]. MRI's role in preoperative planning is crucial, as it helps identify cystic components, inflammatory changes, and the tract's course in relation to the parotid gland, which is important for surgical decision-making. Recent studies emphasize MRI's value in preoperative planning and reducing intraoperative uncertainty when dealing with complex cases involving the parotid or facial nerve [1], [2].

The relationship between the facial nerve and the branchial cleft tract remains a significant concern during surgery. In our patient, the tract passed deep to the facial nerve, which required careful dissection to avoid nerve injury. This anatomical complexity is well-documented in recent studies, which found that Type II FBCAs are more likely to involve the facial nerve, especially when the tract passes deep to or between the branches of the nerve [1], [2]. The deep relationship of the tract with the facial nerve increases the risk of facial paralysis, a complication that has been reported in several series. In our case, facial nerve preservation was achieved through meticulous dissection and identification of the nerve, guided by preoperative MRI and intraoperative monitoring. This approach is supported by the findings of previous studies, which emphasized the importance of facial nerve preservation during surgery for these anomalies, especially when the tract is deep or adherent to the nerve [2], [7].

Surgical excision remains the definitive treatment for FBCAs. The goal is complete removal of the tract to prevent recurrence and other complications. Our patient underwent a superficial parotidectomy with en bloc excision of the tract. This approach is consistent with the management strategy described in other studies, where complete excision of the fistula in patients with recurrent infections was performed [5]. Studies have shown that incomplete excision, especially during the acute inflammatory phase, increases the risk of recurrence and facial nerve damage [6], [7]. The decision to perform a superficial parotidectomy, as recommended in recent case series, is particularly important in Type II FBCAs, where the tract often involves the parotid gland and facial nerve [8].

The postoperative recovery in our patient was uneventful, with no facial weakness or recurrence at 12 months of follow-up. This outcome is consistent with the results reported in other studies, where early surgical intervention and complete excision led to favorable outcomes [1]. However, recurrence and temporary facial paralysis remain possible complications, particularly in cases with prior surgery or delayed intervention, as described in various reports [1], [7]. These complications highlight the importance of early diagnosis and referral to specialized centers to minimize the risk of poor outcomes.

FBCAs present with a wide range of clinical manifestations, from recurrent infection to facial deformities. Imaging, particularly MRI, plays a crucial role in preoperative assessment, while careful surgical planning and facial nerve preservation are key to successful outcomes. Our case highlights the importance of early and accurate diagnosis, complete surgical excision, and careful dissection to prevent recurrence and facial nerve injury. These findings align with the literature, underscoring the need for specialized management of these complex congenital anomalies.

4. Conclusion

First branchial cleft anomalies, though rare, present significant diagnostic and surgical challenges due to their complex anatomical relationships, especially with the facial nerve and parotid gland. Early diagnosis, aided by imaging modalities such as ultrasound and MRI, plays a pivotal role in guiding surgical intervention. Our case highlights the importance of a comprehensive diagnostic approach, including high-resolution imaging, to accurately map the lesion and its relationship with surrounding structures. Surgical management, specifically complete excision with facial nerve preservation, remains the gold standard for treatment, and our favorable outcome underscores the importance of timely and specialized care. The findings in this case align with recent literature, emphasizing the necessity of careful preoperative planning, meticulous dissection, and the role of advanced imaging techniques to optimize patient outcomes and minimize complications such as facial nerve injury or recurrence. As demonstrated, early referral to centers with expertise in managing these anomalies significantly reduces the risk of poor outcomes and enhances the quality of care for patients with first branchial cleft anomalies.

Compliance with ethical standards

Acknowledgments

The authors thank the Department of Otolaryngology – Head and Neck Surgery, Mohammed VI University Hospital Center, Marrakech, for their support in the clinical management of this case.

Disclosure of conflict of interest

The authors declare that they have no conflicts of interest to disclose.

Statement of ethical approval

If studies involve use of animal/human subject, authors must give appropriate statement of ethical approval. If not applicable then mention 'The present research work does not contain any studies performed on animals/humans subjects by any of the authors'

Statement of informed consent

Written informed consent was obtained from the patient for the publication of this case report and accompanying images.

Research involving human participants

This case report was conducted in accordance with the principles outlined in the Declaration of Helsinki. In line within situational policy, ethical committee approval was not required as this report involves a retrospective description of a single clinical case with no identifiable personal information and no experimental intervention.

Consent for Publication

Consent for publication of the case details and figures were obtained from the patient's legal guardian.

Availability of Data and Materials

All data generated or analyzed during this study are included in this published article. Further information is available from the corresponding author upon reasonable request.

Funding

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

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