**Case Report**

# Appropriate imaging in the management of first branchial cleft anomalies

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## ABSTRACT

**Background:** First branchial cleft anomalies (FBCA) are rare clinical entities of the head and neck. The low incidence and varied presentation often result in misdiagnosis and inappropriate treatment. Correct diagnosis is essential for proper management, while an incorrect diagnosis will often lead to inadequate treatment. A good understanding of the regional anatomy and embryology can lead to an early diagnosis and thereby effective management of FBCA. **Purpose:** To present how to diagnose and manage FBCA. **Case report:** A case of a 6-year-old female who had an FBCA with a history of swelling and recurrent discharge from the fistula in the infra-auricular area. Complete excision of the tract was performed without facial nerve complication. **Clinical question:** What is the appropriate imaging for diagnosis FBCA? **Review method:** Scoping review was done to identify the scientific evidence about imaging for diagnosis FBCA. Systemic searching in 4 databases (PubMed, Embase, Proquest, and Web of Science) using keywords "first branchial cleft", "anomaly", and "imaging". **Result:** Three article was found relevant with the topic of imaging for diagnosis FBCA. **Conclusion:** Proper diagnosis of FBCA can lead to proper management and good results. Imaging can provide an anatomical picture of each branchial arch anomaly, which can be very helpful in preoperative planning to determine a definitive surgical approach. Early management of FBCA can reduce the recurrence rate significantly.

**Keywords:** first branchial cleft, anomaly, children, imaging

## *ABSTRAK*

***Latar belakang:*** *Anomali celah brankial pertama (ACBP) adalah entitas klinis yang jarang terjadi pada area kepala dan leher. Insiden yang rendah dan manifestasi yang bervariasi sering mengakibatkan kesalahan diagnosis dan penatalaksanaan yang tidak tepat. Diagnosis yang akurat sangat penting untuk penatalaksanaan yang tepat, sementara diagnosis yang salah seringkali menyebabkan penatalaksanaan yang tidak adekuat. Pemahaman yang baik tentang anatomi dan embriologi dapat mengarahkan pada diagnosis dini, dan dengan demikian penatalaksanaan ACBP menjadi efektif.* ***Tujuan:*** *Untuk menyajikan langkah diagnosis dan manajemen ACBP.* ***Laporan kasus:*** *Seorang anak perempuan berusia 6 tahun dengan* FBCA, *yang memiliki riwayat pembengkakan dan keluarnya cairan berulang dari fistula di daerah infra-auricular. Eksisi lengkap fistula telah dilakukan tanpa komplikasi pada saraf wajah.* ***Pertanyaan klinis:*** *Pencitraan apa yang tepat untuk diagnosis ACBP?* ***Tinjauan literatur:*** *Dilakukan metode* 'scoping review' *untuk mengidentifikasi bukti ilmiah tentang pilihan pencitraan untuk diagnosis ACBP. Dilakukan pencarian literatur melalui* PubMed, Embase, Proquest, *dan* Web of Science, *menggunakan kata kunci* "first branchial cleft", "anomaly", *dan* "imaging". ***Hasil:*** *Ditemukan 3 artikel yang sesuai dengan topik pencitraan untuk diagnosis ACBP.* ***Kesimpulan:*** *Diagnosis ACBP yang tepat membuat tatalaksana ACBP menjadi tepat, dengan hasil yang baik. Pencitraan dapat memberikan gambaran anatomi dari anomali arkus brankial, yang akan sangat membantu dalam perencanaan pra operasi dalam menentukan pendekatan bedah definitif. Manajemen ACBP yang dini dan tepat bisa mengurangi tingkat kekambuhan secara signifikan.*

***Kata kunci:*** *celah brankial pertama, anomal*i, children, *pencitraan*

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## INTRODUCTION

The first branchial cleft anomalies (FBCA) are extremely rare anomalies of the head and neck. FBCA are rare causes of parotid swellings, and comprise less than 1% of all branchial anomalies. They have an estimated incidence of 1 in 100,000 population per year, and are commonly misdiagnosed as preauricular pits, benign parotid cysts, or infected lymph nodes. These anomalies are due to incomplete fusion of the ventral portion of the first and second arches during weeks 4 to 7 of fetal life. During development, the closure time of the cleft is concurrent with the migration of the facial nerve and the emergence of the developing parotid gland, which originates from the second branchial arch; thus, FBCA has a close relationship between these structures. The fissure disappears ventrally or dorsally, and lesions occur in the auricular and parotid regions more often than in the hyoid region.[1-3]

Several classifications of these anomalies have been proposed to facilitate clinical diagnosis. Work[4] in 1972 classified FBCA into two types based on clinical features and histopathology. Type I anomalies are purely ectodermal origin and often present as an epidermal cyst or fistula located in the periaural region with a fistulous tract parallelto the external auditory canal (EAC) that ends in a dead-end at the level of the mesotympanum. These occur superficial to the facial nerve, adjacent to the pinna, and often extend into the postauricular crease. Pathology shows squamous epithelium, but no cartilage or adnexa of the skin. Type II anomalies are ectodermal and mesodermal origin, and present as cysts (closed sacs without openings), sinuses (duct with one opening), or fistula (duct with two openings). They may pass through the parotid gland either superficially, or deep to the facial nerve, and the tract may either terminate in the cartilaginous EAC (perhaps connected to the eardrum), or extend to the face or upper neck. Histology shows squamous epithelium with cartilaginous or skin adnexa.[2-4]

The common clinical presentation is swelling in the periauricular region (24%), parotid (35%), or cervical region (41%), with possible external drainage from a skin pit due to repeated episodes of infection or epithelial desquamation. The rare incidence and variation of clinical features often make diagnosis difficult. It is important to consider this diagnosis in all cases with swelling or infection of the external ear, including the presence of cysts, sinuses, or fistula in the region of the angle of the mandible.[3,5]

Radio-diagnostic/imaging usually is needed for diagnosis. Although, with the aid of radiological images, they still can be easily misdiagnosed, leading to improper treatment such as incision and drainage with high recurrence risk. The only definitive treatment of FBCA is total excision of the lesion. FBCA is frequently related to the facial nerve, which is susceptible to damage during surgery, causing post-operative facial palsy and affecting the quality of life in children. Therefore, the management of FBCA is quite challenging.[1,3,5]

This case report was presented to review the diagnosis and management of the first branchial cleft anomalies.

## CASE REPORT

A 6 years old girl presented to us with intermittent swelling and discharge from an opening under her right ear since she was 1.5

years old. She had a history of a mass under her right ear, which had an opening that outflowing purulent discharge. She had been diagnosed with an abscess and had undergone incision drainage 2 years previously, but the intermittent swelling and discharge persisted. Another surgery was conducted, diagnosed as a pre-auricular fistula. But the symptoms recurred within 3 months later.

From the physical examination, general condition of the patient was good with normal vital signs. Otomicroscopy examination showed wide ear canal, no secrete and tympanic membrane was intact in both ears. Two small openings were found under her right ear, with a scar mark from previous surgery without sign of acute infection (Fig.1).

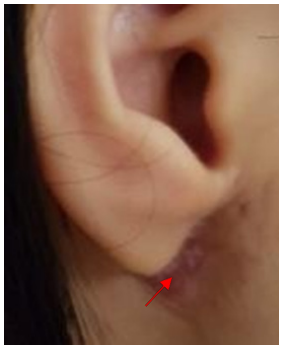

**Figure 1. Before surgery**

On fistulography, the contrast was injected through the external fistula opening at the right infra-auricular region. It showed cutaneous-subcutaneous fistula at the right infra-auricular region, with pouch formation at the mid and distal area with the size of 0.4 x 1.2 cm. There was no distal fistula contrast flow from the pouch to other structures. (Fig. 2)

The MRI examination showed fistula formation starting from the right infra-posterior parotid gland, heading to supero-lateral, then turning infero-posterior to the right infra-auricular region (right infra pinna, at the area where the marker was attached). The fistula tract also had a small blind-ended branch at the base, which directed posteriorly(± 1-2 mm). The fistula tract to the external ear canal was obscure. The medial aspect of the infra pinna cutaneous opening of the fistula showed a granulation process and cyst-like formation ±5 mm. These features could be suspected as first branchial cleft cyst Type 1. Subcutaneous wall thickening of the external auditory canal was caused by a chronic inflammatory reaction. (Fig.3)

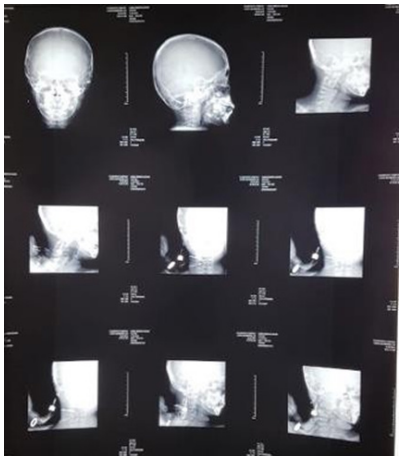

**Figure 2. Fistulography**

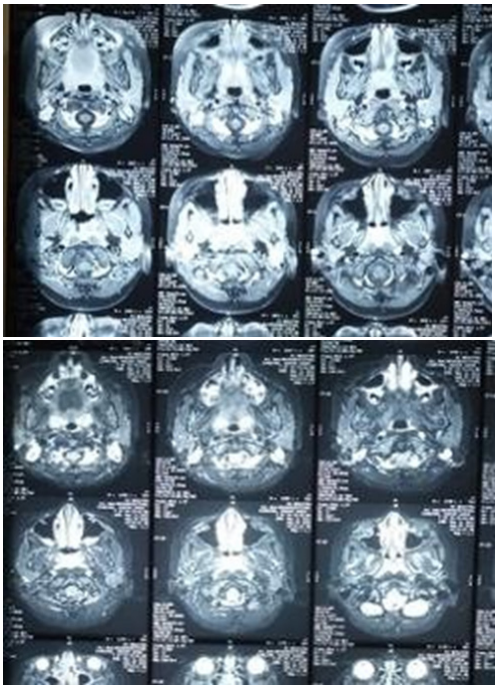

**Figure 3. MRI**

We diagnosed the patient with Type I FBCA and decided to excise the tract without parotidectomy. A sinusectomy under general anesthesia was performed guided by injected methylene blue into the external sinus opening. Sinus tract dissection was performed infra auricularly. Afterwards, the incision was then extended to the inferior area of the tragus, up to 2-3 cm under the lobule. All tracts were explored and sacs were removed completely along with the surrounding tissues (Fig. 4).

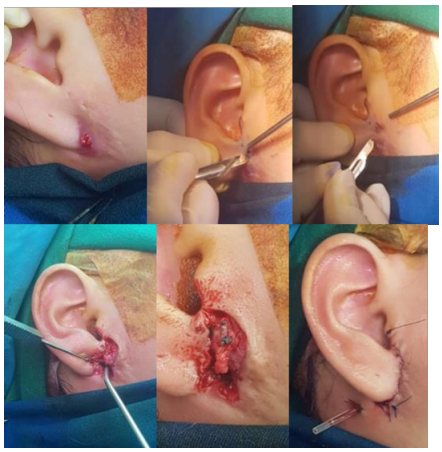

**Figure 4. Intraoperative**

Histopathologic examination result was in accordance with an epidermal cyst, thus confirming the diagnosis of Type I FBCA. There were no complications, and the patient recovered with no signs of recurrence until 6 months after surgery (Fig. 5-6).

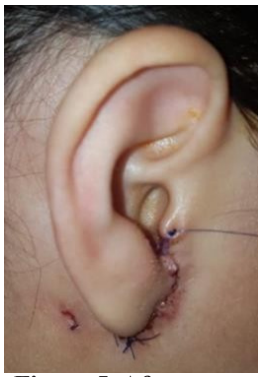

**Figure 5. After surgery day-2**

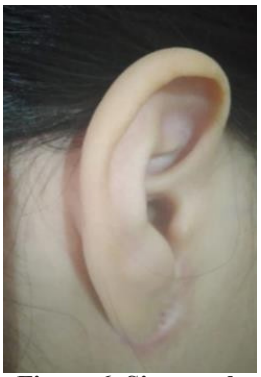

**Figure 6. Six month after surgery**

## CLINICAL QUESTION

Based on the presented case, the background question proposed was: "What is the appropriate imaging for diagnosis FBCA?"

Patient: Patient with FBCA.

Concept: Imaging tools for diagnosing FBCA.

Context: General setting.

Inclusion criteria: case report, case controland, cohort study design.

## REVIEW METHOD

A comprehensive and systematic literature search was conducted using various electronic databases including PubMed, Embase, Proquest, and Web of Science. The search was performed using a combination ofkeywords and Medical Subject Headings (MeSH) terms, and a Boolean search system was employed to combine and analyze the terms effectively. The search terms used keywords included "first branchial cleft", "anomaly", and "imaging". Non-English articles were excluded.

## RESULT

The literature searching used eligibility criteria based on keywords from clinical questions, found three articles which was relevant to our clinical question.

## DISCUSSION

The symptoms of FBCA usually begin early in life, either in infancy or childhood, but diagnosis and appropriate surgical treatment are often delayed for years, in some cases into adulthood. This anomaly is easily misdiagnosed. Triglia et al.[6] reported a delay of 3.5 years between the onset of the

first clinical symptom and when adequate treatment was implemented, with almost 50% of patients not receiving successful treatment.[2,6,7]

Clinical manifestations of FBCA include repeated swelling or discharge of the skin fistula in Pochet's triangle area. This triangle is bounded by the external ear canal superiorly, the mental region anteriorly and the hyoid bone inferiorly. Swelling could be mistaken as other cervical mass. In children with chronic or recurrent upper neck infections, especially in the setting of ipsilateral ear disease, attending physicians should seriously consider the possibility that the patient might have an FBCA. The common otological manifestation is otorrhea or recurrent otitis externa, with infective exacerbation. Along with this, there might be complaints of episodic swelling draining into the external auditory canal. Some patients complain of hearing loss and sensation of stuffiness in the ear, either due to obstruction of the ear canal or due to edema associated with otitis externa. Rarely, does the first branchial cleft sinus present with cholesteatoma. Some pathognomonic of FBCA include a history of multiple incisions and drainage procedures for an abscess in the upper neck region. Shinn et al.[8] noted that more than half of the children had received an incision and drainage before the surgical removal of the fistula, and more than a quarter of the subjects had a history of surgery.[1-3,8]

This case report described a girl with repeated swelling and discharge from an opening under her right ear. A recurrent infection occurred when she was 1.5 years old. There was no history of ear discharge, hearing loss nor facial asymmetry. She also had a history of multiple surgical procedures before she came to us.

Complete history taking, and head-neck examination are mandatory in all cases. Pre-operative diagnosis is challenging and misdiagnosis is not uncommon. FBCA should be considered as a differential diagnosis in children complaining of any openings, swelling, inflammation, persistent cyst or fistula in the neck at the level I, IIa/IIb, on the face, or postauricular area, accompanied with supporting history and clinical examination. A thorough otoscopic examination should be performed, preferably using binocular microscopy. The structure of the external ear canal should be examined to identify a possible fistula, or asymptomatic adhesion between the external ear canal and the tympanic membrane, granulation tissue, or middle ear disease.[3,8]

The radiologic examination also plays an important role in diagnosing these anomalies. It allows anatomic imaging of the branchial arch anomaly, which helps preoperative planning of the definitive surgical approach. On imaging, first branchial sinuses are related to the parotid gland and/or the lower margin of the pinna. The diagnosis is established if a tract directed toward the external auditory canal could be identified. Several techniques can be used, including fistulography, ultrasonography, CTscan, MRI, and CT fistulography.[2,9]

Pre-operative fistulography is used to visualize the external and internal openings as well as the course of a sinus or fistula, and it is performed to find out the extent of the tract, but it cannot demonstrate its anatomical relationship to the surrounding normal structures. Other modalities have been recommended to help confirm the diagnosis of FBCA, such as CT scan and MRI.[9,10]

Radiation exposure is a concern for children, therefore USG can be the first choice in helping the diagnosis of head and neck tumors in young ages. In uncomplicated anomaly, USG shows a centrally hypoechoic appearance with posterior acoustic enhancement, a typical cyst characteristic. Variations include septated cysts, inhomogenous, or even solid appearance.[11,12]

Investigations such as CT scans and MRIs provide the best picture for these cases. This type of imaging help to describe the size of the lesion and the anatomical relationship of FBCA to important surrounding structures, such as the facial nerve, external ear canal, and parotid gland. The CT scan could show the relationship of the lesion to the outer ear canal and the middle ear. MRI plays a crucial role in diagnosis and differentiation of this entity from other cystic lesions of the parotid gland. MRI provides accurate information on the extent/course of the lesion, especially in the parotid area, and the cyst wall can be clearly identified on contrast-enhanced MRI. In Type 1, a cystic mass appears around the auricle; and in Type 2, it extends from the external ear canal to the angle of the mandible. The examination results show a fluid-filled cyst, with or without signs of infection or inflammation, depending on the timing of the scan with varying thickness of the cyst wall. On CT imaging, cystic mass could be presented as superficial/between/deep to the parotid salivary gland.[3,5,9,11]

CT fistulography is a relatively new technique, where contrast is injected into the tract during examination. This allows better visualization of the fistula tract and its relation to surrounding structures compared to conventional CT.[10,12]

After her second surgery, our patient underwent MRI which showed a Type I first branchial cleft anomaly. Fistulography was also performed to find out the extent of the tract.

Successful treatment necessitates a thorough understanding of the complex embryology involved in the pathogenesis of these anomalies, the close relationship of the branchial anomaly to the facial nerve, and a wide range of clinical presentations of such branchial anomalies. The relation between facial nerve and the sinus or fistulous tract seems to be variable. The course of fistulous tracts to the facial nerve could be lateral (41%), medial (37%), or between the branches (22%); whereas the sinus tract usually has a superficial course.[2,13] Guo and Guo[13] suggested that the most appropriate time for surgery is after 4 years old. At this age, the facial nerve has fully developed, making surgery easier, and reducing the risk of facial nerve damage.[1,13]

The principles of FBCA management are early diagnosis, infection control, and complete surgical excision with preservation of the facial nerve. Spontaneous regression of FBCA is unlikely, and recurrent infections are common, therefore surgery is the best therapy. Most patients underwent incision and drainage procedures before the definitive surgery is performed. The absence of previous incision and drainage procedures is ideal at the time of the definitive FBCA surgery. Previous drainage or cyst rupture may lead to scar tissue and fistula formation. Definitive surgery should be performed after the acute infection has resolved, and complete antibiotics administration. However, for persistent symptoms such as swelling and pain, this period can be shortened. Any scar or fistula should be removed along with the tract and cyst.[3,8,9]

Surgical methods vary from complete resection preserving the normal structure, to a superficial parotidectomy approach which carries the risk of damage to the facial nerve and, if necessary, reconstruction of the ear structures. The surgical technique approach depending on the type of lesion location. In Type 1, where the lesion is located superficial to the facial nerve, identification of the nerve is not necessary. Superficial parotidectomy is the preferred procedure for deep-seated lesions, or between the facial nerves, which typically occur in Type 2. The facial nerve should be fully visualized to ensure nerve safety. The deeper the position of the lesion, the greater the amount of tissue that should be removed in a superficial parotidectomy.[9,13]

One of the keys to complete excision is keeping the tract, cyst, and any fistula or scar tissue intact. A lacrimal probe can be used throughout the case to ensure that the tract does not get disrupted and to determine the orientation of the tract. A dye such as methylene blue or gentian violet can be infused into the tract to help identify and distinguish the abnormal tract from normal tissue. In many cases, it is advisable to remove structures closely related to the cyst or tract, such as resection of skin, cartilage of the external ear canal, or posterior mandibula to prevent recurrence.[3,13]

Shin et al.[8] also described the algorithm for the management of FBCA in their literature. If the child has a Work type I anomaly as indicated by a pit or cyst around the lobule, imaging may not be necessary, although preferences will likely be institutional. Surgical excision of the tract, superficial to the facial nerve, is performed by a ''mini-facelift'' incision behind the tragus and around the lobule with facial nerve monitoring (without parotidectomy). If a connection or cyst is visible on the tympanic membrane or external ear canal, then a simultaneous tympanoplasty or canaloplasty should be performed. If a Work type II anomaly is suspected, imaging should be performed to facilitate surgical planning and to identify the location of the tract that lies to the facial nerve. A parotidectomy approach with the initial identification of the facial nerve and excision of the entire tract is indicated. Simultaneous tympanoplasty or canaloplasty should be performed if a connection between the superficial skin and external ear canal or tympanic membrane is identified, or if a cyst involves the external ear. Facial nerve monitoring should be considered in all cases.[8]

In our patient, the lesions lay superficial to the facial nerve, and we did not need to identify the nerve during surgery. In patients with Type 1 FBCA, total methylene blue-guided fistulectomy without parotidectomy could be chosen. An elliptical incision is made around the puncta. Blunt dissection is then performed to extend the incision and allow complete removal the of fistula.

In the presented case, the histopathologic examination showed an epidermal cyst, no adnexa, nor cartilage. In Type 1 FBCA, the histopathologic features are keratin-filled cysts lined with stratified squamous epithelial or ciliated columnar epithelial, due to their ectodermal origin.[1,3]

In conclusion, FBCA is a commonly misdiagnosed rare entity. Clinical manifestations include recurrent swelling or discharge from the fistula in the periauricular or Pochet's triangle region. Comprehensive knowledge regarding complex embryology involved in the pathogenesis, location concerning facial nerve, and clinical features of the anomaly, is crucial for successful treatment. A radiologic examination is important in confirming the diagnosis of the anomaly, which also leads to proper management. Total tract excision is the definitive treatment, with careful pre-operative planning purposely to preserve the facial nerve during resection.

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