First Branchial Cleft Anomalies: Otologic Manifestations and Treatment Outcomes

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Sponsorships or competing interests that may be relevant to content are disclosed at the end of this article.

Abstract

Objective. This study describes the presentation of first branchial cleft anomalies and compares outcomes of first branchial cleft with other branchial cleft anomalies with attention to otologic findings.

Study Design. Case series with chart review.

Setting. Pediatric tertiary care facility.

Methods. Surgical databases were queried to identify children with branchial cleft anomalies. Descriptive analysis defined sample characteristics. Risk estimates were calculated using Fisher's exact test.

Results. Queries identified 126 subjects: 27 (21.4%) had first branchial cleft anomalies, 80 (63.4%) had second, and 19 (15.1%) had third or fourth. Children with first anomalies often presented with otologic complications, including otorrhea (22.2%), otitis media (25.9%), and cholesteatoma (14.8%). Of 80 children with second branchial cleft anomalies, only 3 (3.8%) had otitis. Compared with children with second anomalies, children with first anomalies had a greater risk of requiring primary incision and drainage: 16 (59.3%) vs 2 (2.5%) (relative risk [RR], 3.5; 95% confidence interval [CI], 2.4-5; \( P < .0001 \)). They were more likely to have persistent disease after primary excision: 7 (25.9%) vs 2 (2.5%) (RR, 3; 95% CI, 1.9-5; \( P = .0125 \)). They were more likely to undergo additional surgery: 8 (29.6%) vs 3 (11.1%) (RR, 2.9; 95% CI, 1.8-4.7; \( P = .0025 \)). Of 7 persistent first anomalies, 6 (85.7%) were medial to the facial nerve, and 4 (57.1%) required ear-specific surgery for management.

Conclusions. Children with first branchial cleft anomalies often present with otologic complaints. They are at increased risk of persistent disease, particularly if anomalies lie medial to the facial nerve. They may require ear-specific surgery such as tympanoplasty.

Keywords

branchial cleft, otitis, congenital neck mass, pediatric otolaryngology
source, the primary objective of the current study was to describe the presentation of first BCAs at our institution, with attention to otologic manifestations of disease. A second objective was to compare treatment outcomes of children with first BCAs to children with other types of BCAs to determine the extent to which children with first anomalies are at greater risk of delayed diagnosis or persistent disease. Our hypothesis was that children with persistent disease would often require ear-specific surgery for definitive management.

**Methods**

This investigation is a case series that received institutional review board approval from Seattle Children’s Hospital, a pediatric tertiary care facility. All children and adolescents who were surgically treated for congenital neck masses between 2004 and 2013 were identified through query of the facility’s surgical database using the following Current Procedural Terminology (CPT) codes: 21556, 21557, 38510, 42408, 42815, 60280, and 60281.

A retrospective chart review was performed to identify subjects from age birth to 21 years who were treated for BCA. Diagnosis of branchial anomaly was made on the basis of clinical evaluation, imaging characteristics, and surgical pathology.

First BCAs were defined by a pit or cyst identified in level I or level II or the postauricular region with imaging and surgical confirmation of a tract leading toward the ear canal. Second branchial cleft sinuses and fistulae were defined by a congenital pit along the anterior border of the sternocleidomastoid muscle with surgical confirmation of a tract heading partially or completely to the tonsillar fossa. Second branchial cleft cysts were defined by an isolated level II cyst lined by ciliated columnar epithelium. Third or fourth branchial anomalies were defined by level III or level IV neck inflammation, thyroid involvement, and/or a pit in the ipsilateral piriform sinus. In all cases, diagnosis of branchial cleft anomaly was confirmed by review of final pathology reports.

Data were collected regarding demographic characteristics, presenting signs and symptoms, procedures performed, and treatment outcomes. Clinical records of children with first and second BCAs were reviewed with attention to otologic manifestations of disease.

**Statistical Analysis**

Univariate analyses were carried out to obtain descriptive statistics such as means, confidence intervals, and frequencies for both groups. To make comparisons between the groups, we performed inferential testing using one-way analysis of variance. Risk estimates were obtained through calculation of risk ratios using Fisher’s exact test. $P < 0.05$ was considered statistically significant. Stata 13.1 (StataCorp, College Station, Texas) statistical software was used for all analyses.

**Results**

Review identified 126 patients with BCAs. Of these, 27 (21.4%) had first BCAs, 80 (63.4%) had second BCAs, and 19 (15.1%) had third or fourth branchial anomalies.

Clinical characteristics and presenting symptoms of the subjects are summarized in Table 1. Subjects with first BCAs were more likely to be female (63%) and to have left-sided lesions (63%). Patients with second BCAs were more likely to have right-sided lesions (63.7%), while patients with third or fourth BCAs had predominantly left-sided lesions (89.5%).

While most children were correctly diagnosed with BCA, chart review also identified a subgroup of patients who did not have BCA but rather the more commonly encountered entity of preauricular pits. These children most commonly presented with infected pits or abscesses anterior to the tragus or helical root; these lesions were identified on the right, left, or bilaterally nearly in equal numbers (5 bilaterally, 4 on the left, and 6 on the right). Findings during surgical excision included a pit, mass, or skin tag anterior to the tragus or helical root in all cases.

Children with first and second BCAs had similar age at diagnosis, 2.08 years versus 1.51 years ($P = 0.4$, not significant). In contrast, children with third or fourth BCAs were significantly older with a mean age of 6.37 years ($P < 0.001$). Children with first BCAs most commonly presented with otologic complaints (40.7%), while those with second BCAs were most likely to present with draining sinus or fistulae (62.5%). All children with third or fourth BCAs presented with neck mass or abscess.

**Table 1. Characteristics of Different Types of Branchial Cleft Anomalies.**

<table>
<thead>
<tr>
<th>Type of Anomaly</th>
<th>No.</th>
<th>Age at Diagnosis, Mean (95% CI), y</th>
<th>Female Sex, No. (%)</th>
<th>Laterality, No. (%)</th>
<th>Presenting Symptom, No. (%)</th>
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</thead>
<tbody>
<tr>
<td>First</td>
<td>27</td>
<td>2.08 (1.07-3.08)</td>
<td>17 (63)</td>
<td>17 (63)</td>
<td>10 (37)</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>6 (22.2)</td>
</tr>
<tr>
<td>Second</td>
<td>80</td>
<td>1.51 (0.67-2.36)</td>
<td>34 (42.5)</td>
<td>24 (30)</td>
<td>51 (63.7)</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>5 (6.3)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>50 (62.5)</td>
</tr>
<tr>
<td>Third</td>
<td>19</td>
<td>6.37 (4.12-8.62)</td>
<td>10 (52.6)</td>
<td>17 (89.5)</td>
<td>2 (10.5)</td>
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Abbreviation: CI, confidence interval.

**Table 2** lists otologic examination findings of the subjects. Although a small number of patients with second BCAs had otitis media or EAC deformity, an otologic problem was not given as the primary complaint for any of these...
patients. In contrast, patients with first BCAs had a variety of otologic complications, ranging from EAC cyst to cholesteatoma. Even if not the presenting complaint, the presence of otitis media was noted from clinical examination records. Seven of 27 children with first BCAs and 30 of 80 children with second BCAs were noted to have otitis media (relative risk [RR], 2.8; 95% confidence interval [CI], 1.6-4.7; \( P = .006 \)). Among the 7 children with first BCAs who had otitis, 4 had a history of acute otitis media, while 3 had a history of chronic otitis media with effusion. None had TM perforation.

An EAC web, or abnormal connection between the anterior-inferior or posterior-inferior canal wall and TM, can signal the presence of a first branchial cleft anomaly; 6 of our subjects (22.2%) were discovered to have such a web on evaluation. In addition, children with first BCAs displayed a range of other EAC deformities: 5 (18.5%) had an EAC mass or cyst, while 5 (18.5%) had EAC duplication. In comparison, 3 (3%) of the children with second BCAs displayed EAC deformities, which were associated with hemifacial microsomia or Down syndrome.

Table 3 contains a comparison of outcomes among patients with first and second BCAs. Compared with children with second BCAs, children with first BCAs had a greater incidence of primary incision and drainage: 16 (59.3%) vs 2 (2.5%) (RR, 3.5; 95% CI, 2.4-5; \( P < .0001 \)). Of the 16 children with first BCAs who underwent primary incision and drainage, 11 presented with an acute abscess, while 5 children presented with a draining pit. After resolution of their acute infection, the children underwent primary excision. Compared with children with second BCAs, children with first BCAs were more likely to have persistent disease following primary excision: 7 (25.9%) vs 2 (3.8%) (RR, 3; 95% CI, 1.9-5; \( P = .0025 \)). They were also more likely to undergo additional surgery: 8 (29.6%) vs 3 (11.1%) (RR, 2.9; 95% CI, 1.8-4.7; \( P = .0025 \)). There was not a significant difference in rates of persistent disease between children with first and children with third or fourth BCAs. Of the 19 children with third or fourth BCAs, 11 (57.9%) underwent incision and drainage prior to excision, 7 (36.8%) experienced persistent disease, and 7 (36.8%) required additional surgery.

Length of follow-up period was not significantly different among the anomaly types. Children with first BCAs had a mean follow-up period of 16 months (95% CI, 6-27 months), children with second BCAs had a mean follow-up period of 8 months (95% CI, 4-12 months), and those with third or fourth anomalies had a follow-up of 11 months (95% CI, 1-21 months).

Table 4 describes the surgical management of the 27 patients with first BCAs. Thirteen subjects (48.1%) underwent...
superficial parotidectomy on primary excision, with 4 of these patients also undergoing tympanoplasty or tympanomastoidectomy. One patient underwent tympanomastoidectomy alone after the identification of an EAC duplication, a type of first branchial cleft anomaly. The duplication was located superficial to the facial nerve. The remainder of children underwent simple excision at the time of primary surgery. Among the 7 patients who required revision surgery, more than half (57.1%) underwent otologic surgery, either tympanoplasty or tympanomastoidectomy. There were 9 patients (33.3%) who had Work type II anomalies that coursed medial to the facial nerve; all eventually required parotidectomy either at primary or repeat excision. Of the 7 patients who required additional surgery, 6 (85.7%) had anomalies medial to the facial nerve.

Facial nerve complications were also evaluated among the patients with first BCAs. Only 1 patient (3.7%) had confirmation of permanent facial nerve weakness, an adolescent who presented to our facility with facial nerve palsy after undergoing multiple procedures at outside facilities. In addition, 3 patients (11.1%) developed temporary weakness of the marginal mandibular nerve that completely resolved, while 2 (7.4%) patients experienced weakness of unknown duration due to loss to follow-up. Among the 5 cases that occurred at our institution, the first branchial cleft anomaly was identified medial to the facial nerve during the excision procedure.

Discussion

Children with first BCAs often undergo multiple procedures to treat infectious complications and ultimately remove these lesions. In the current study, more than half of the children were treated with incision and drainage prior to excision, and over a quarter required repeated excision procedures. Risk of persistent disease and repeat surgery were much higher for these children than for children with second BCAs. This indicates that the diagnosis of first BCA may not be recognized until the anomaly is infected and requires incision and drainage. Interestingly, our findings suggest that children with third or fourth branchial anomalies also have high rates of persistent disease, which is consistent with previously published literature\(^1\) and should be explored further in future investigations.

Careful otologic evaluation can assist providers in making correct diagnosis of first BCAs\(^1\). Previous case series have described otologic complications, including cholesteatoma\(^1\); however, we were surprised by the high frequency of children who presented with a primary otologic complaint. Otologic complications were the most frequent presenting symptoms, occurring in nearly half of the cases. Otorrhea and otitis media were identified in about one-fourth of the cases, and 15% of patients were found to have cholesteatoma. Seven children with first BCAs had otitis media, of whom 4 had a history of acute otitis media and 3 had a history of chronic otitis media with effusion. None of these children had tympanic membrane perforation. In addition, more than half of the patients were noted to have an EAC anatomical deformity.

Perhaps the most complicated case encountered was a 2-year-old male who presented with a left level II neck abscess and purulent otorrhea. The patient was ultimately diagnosed with both Work type I and Work type II anomalies. During the child’s initial incision and drainage procedure, pressure on the neck abscess resulted in copious expression of pus from the ear canal (Figure 1), confirming the diagnosis of a Work type II anomaly. After antibiotic therapy, the patient had persistent erythema and drainage at the original incision site and in the postlobular crease (Figure 2). Otoscopy revealed both anterior-inferior and posterior-inferior TM webs along with some persistent granulation at the anterior inferior sulcus (Figure 3). The 2 first branchial anomalies were then excised via a superficial parotidectomy approach with facial nerve dissection (Figure 4). The proximal cartilaginous portion of the main fistula, which ran deep to the facial nerve, was shaved off at the bony cartilaginous junction of the anterior ear canal. The patient experienced continued wound infection at the incision area directly inferior to the lobule. Cultures grew *Pseudomonas*, indicating a likely continued connection to the ear canal or middle ear. Otoscopy demonstrated a persistent area of granulation at the point of origin of the anterior web at the anterior sulcus. A tympanoplasty was performed with excision of the anterior-inferior TM, curettage of the pit at the anterior sulcus, and reconstruction with fascia. The infection resolved, and the ear canal and tympanic membrane healed (Figure 5). This is a rare case of 2 first branchial anomalies in 1 patient and emphasizes the point that Work type II fistulas can present at a young age with a sinus tract running deep to the facial nerve. Failure to adequately address the otologic component of the lesion can result in persistent infection, as was the case for this patient.

The approach to a child with a first branchial cleft anomaly lies in accurate diagnosis; Figure 6 contains a potential algorithm for management. First anomalies should be among...
the differential diagnoses considered when children present with a pit or inflammatory response in neck level I, high in level IIa/IIb, on the face, or in the postauricular area. A thorough otoscopic examination is indicated, with the use of binocular microscopy if available. The examiner should look specifically for a canal web, granulation tissue, or middle ear disease. Evidence of purulent expression from the ear with simultaneous pressure on the neck or infected lesion indicates a communication between the lesion and the EAC. Imaging, either with contrast computed tomography (CT) or magnetic resonance imaging (MRI), can help clarify the diagnosis. If a child presents with acute infection, incision and drainage and antibiotics are indicated prior to definitive excision.

When deciding the optimal approach for primary excision, patients who present with a pit or abscess anterior to the angle of mandible, at cervical level I or II, are more likely to have an anomaly deep or medial to the facial nerve (Work type II). In our study, all of these patients required parotidectomy, and many required otologic surgery. If there is any suspicion of Work type II anomaly, then imaging should be performed to assist with operative planning and to determine where the tract lies in relation to the facial nerve. A parotidectomy approach with preliminary identification of the facial nerve and excision of the entire tract is indicated. Concurrent tympanoplasty or canalplasty should be performed if a connection between the superficial skin and EAC or TM is seen or if a cyst involves the external ear. Facial nerve monitoring should be considered in all cases. In the current study, all patients with facial nerve weakness had a Work type II anomaly medial to the nerve.

If the child has a Work type I anomaly, indicated by a pit or cyst around the lobule, then imaging may not be...
required, although preferences will likely be institution specific. Surgical excision of the tract, superficial to the facial nerve, is performed with a “mini-facelift” incision behind the tragus and around the lobule with facial nerve monitoring. If there is a visible connection or cyst on the tympanic membrane or external auditory canal, then concurrent tympanoplasty or canalplasty should be performed.

Children with preauricular pits may be misdiagnosed as having a first BCA; however, we found these children to always present with a lesion near the helical root. These children most commonly presented with infected pits or abscesses and were identified on the right, left, or bilaterally nearly in equal numbers. Preauricular pits are a distinct diagnosis and should not be confused with BCA. They occur anterior to the tragus or helical root, often become infected, and do not involve the facial nerve. Imaging prior to surgical excision is often not necessary.

As a retrospective study, this investigation has a number of limitations, including incomplete records, loss to follow-up, and potential misclassification of anomaly type. However, the large difference in persistent disease between first and second BCAs strongly suggests that there is a need for improvement in surgical management of first BCAs.

Misdiagnosis and inadequate treatment of first BCAs have the potential to result in recurrent infection, scarring due to multiple procedures, and facial nerve injury. For children with chronic or recurrent upper neck infections, especially in the setting of ipsilateral ear disease, providers should strongly consider the possibility that the subject may have a first branchial cleft anomaly. Children with anomalies that occur medial to the nerve are especially at risk for persistent disease. Preoperative imaging should be performed to define this relationship, so that families can be counseled appropriately. If otologic involvement is suspected, then it should be addressed with otologic surgery at the time of primary excision if possible.

Conclusion
First BCAs are associated with a wide range of otologic manifestations. These children, particularly those with Work type II lesions medial to the facial nerve, experience a relatively high frequency of persistent disease, and otologic surgery may be required for definitive surgical management.

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Author Contributions
Justin R. Shinn, study design, data collection, manuscript preparation; Patricia L. Purcell, study design, data analysis, manuscript revision; David L. Horn, data collection, manuscript revision; Kathleen C. Y. Sie, data collection, manuscript revision; Scott C.
Manning, study conceptualization, study design, data collection, manuscript preparation and revision.

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