First Branchial Cleft Anomalies

A Study of 39 Cases and a Review of the Literature

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Objectives: To identify the clinical and anatomical presentations and to discuss the guidelines for surgical management of anomalies of the first branchial cleft.

Design: Retrospective study.

Setting: Three tertiary care centers.

Patients: Thirty-nine patients with first branchial cleft anomalies operated on between 1980 and 1996.

Intervention: All patients were treated surgically. Complete removal of the lesion required superficial parotidectomy with facial nerve dissection in 36 cases. The relationship of the facial nerve and anomalies is discussed.

Results: Anatomically, 3 types of first branchial cleft anomalies are identified: fistulas (n=11), sinuses (n=20), and cysts (n=8). Clinically, 3 types of presentation are noted: chronic purulent drainage from the ear (n=12), periauricular swelling in the parotid area (n=18), and abscess or persistent fistula in the neck located above a horizontal plane passing through the hyoid bone (n=21). A membranous attachment between the floor of the external auditory canal and the tympanic membrane was observed in 10% of cases. The facial nerve was located lateral to the anomaly in 39% of cases.

Conclusions: Before definitive surgery, many patients (n=17) underwent incision and drainage for infection owing to the difficulties in diagnosing this anomaly. Wide exposure is necessary in most cases, and a standard parotidectomy incision allows adequate exposure of the anomaly and preservation of the facial nerve. Complete removal without complications depends on a good understanding of regional embryogenesis, a knowledge of the circumstances surrounding discovery, an awareness of the different anatomical presentations, and a readiness to identify and protect the facial nerve during resection.


RESULTS

Twenty-seven of the patients were female and 12 were male. The defect was located on the right side in 18 cases and on the left side in 21 cases. Bilateral lesions or a family history suggesting a hereditary origin was not noted. In 3 cases, the lesion was discovered in adult patients aged 23, 42, and 49 years and was removed within 2 months thereafter. In the other cases, initial presentation ranged between birth and 17 years, with a mean age of 2.4 years, while age at surgical resection or persistent fistula in the neck located above a horizontal plane passing through the hyoid bone (n=21). A membranous attachment between the floor of the external auditory canal and the tympanic membrane was observed in 10% of cases. The facial nerve was located lateral to the anomaly in 39% of cases.

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PATIENTS AND METHODS

Between 1980 and 1996, a total of 39 patients underwent surgery for first branchial cleft anomalies at La Timone Hospital in Marseille, France (n=20), the Trousseau Hospital in Paris, France (n=13), and the Albany Medical Center in Albany, NY (n=6). The records of these patients were reviewed to determine the side on which the defect occurred, sex, age at initial presentation, age at the time of surgical management in our departments, and previous surgical procedures.

Clinically, 3 types of clinical manifestations were observed alone or in combination in the same patient. Cervical symptoms consisted of a pit-type depression near the angle of the mandible. Pressing the area produced a small string of clear material. During periods of infection, the discharge could become purulent and lead to submandibular adenitis. Parotid symptoms consisted of small isolated masses located either at the lower pole of the parotid area or against the mastoid area. These tumor-like lesions were generally found because of a rapid increase in size during inflammatory periods. Auricular symptoms consisted of otorrhea with a fairly mucopurulent or purulent discharge from the ear.

Anatomically, anomalies were classified as fistulas (a tract with 2 openings), sinuses (a tract with a single opening), or cysts (a tract with no opening). In patients with fistulas, the lower opening (Figure 1) was located between the sternocleidomastoid muscle in the back, the angle of the mandible in the front, and the hyoid bone below, while the upper opening was located on the floor of the external auditory canal. In patients with sinuses, the opening was located in 1 of these sites. Otoscopy was performed in patients to detect abnormalities of the tympanic membrane or of the middle ear. Special attention was made to detect a membranous attachment between the medial part of the floor of the external auditory canal and the tympanic membrane. This attachment appeared as a triangle with the upper tip extending to the handle of the malleus, the base resting on the external auditory canal, and the medial side pressing against the pars tensa (Figure 2).

The highlights of the surgical procedure were the approach, exposure of the facial nerve, and determination of its relationship with the anomaly. In 26 cases, histological slides were reexamined to check for the presence of squamous epithelium, adnexal skin structures, and cartilage. Excision required wide exposure with parotidectomy and dissection of the facial nerve in 36 cases. The facial nerve was located medial to the anomaly in 18 cases and crossed over it in 14 cases. In 4 cases, the anomaly coursed between the branches of the facial nerve. Reexamination of the 26 available surgical specimens revealed the presence of squamous epithelium in all cases, adnexal skin structures in 18 cases, and cartilage in 13 cases. Paralysis of the inferior branch of the facial nerve occurred in 6 cases: temporary in 5 cases and permanent in 1 case. Recurrence requiring a second surgical procedure occurred in 3 patients, with a mean delay of 11 months (range, 4 months to 2 years). The mean follow-up was 3.7 years from the last intervention, including subsequent procedures. No further manifestation of the anomalies has been observed.

COMMENT

To our knowledge, the present retrospective series of 39 first branchial cleft anomalies is one of the largest published to date. The only series of similar size is that of Olsen et al,1 which included 38 cases collected over a 28-year period. Our findings confirm the likelihood of misdiagnosis and inappropriate treatment. In our pediatric experience, the delay between initial presentation and adequate treatment was 3.5 years, which is comparable to the 4-year delay reported by Ford et al.2 Early diagnosis and management rest on a combined knowledge of regional embryogenesis, the circumstances surrounding discovery, the different anatomical forms, and the consistent role of infection.

Anomalies of the first branchial cleft arise from incomplete closure of the ectodermal portion of the first branchial cleft. Whether the defect is a fistula, sinus, or cyst depends on the degree of closure. The anomaly begins on the floor of the external auditory canal either at the level of the bony-cartilaginous junction or in the cartilaginous portion, follows the seam between the mandibular and hyoid arches, and ends in the submandibular region more or less distally, depending on the extent of the disturbance of the fusion.

In our series, all first branchial cleft anomalies were located within a roughly triangular area (Figure 3). The apex of the triangle was formed by the external auditory attachment between the tympanic membrane and the floor of external auditory canal was observed in 4 patients.
canal medially to the intertragic notch, which is a superficial remnant of the external groove of the first cleft. The base of the triangle was formed by a line between the tip of the chin and the middle of the hyoid bone. The other 2 sides of the triangle were formed by 2 lines, one curving from the external auditory canal to the tip of the chin along the lower edge of the mandible and the other curving from the external auditory canal to the greater horn of the hyoid bone. The anomalies did not extend beyond a horizontal plane passing through the hyoid bone. The incidence of malformation was higher at the top of the triangle, near the external auditory canal and in the parotid region, than at the base of the triangle, in the vicinity of the hyoid bone. Only 8 patients in the present series presented with fistulas or abscesses strictly confined to the lower part of the triangle (Figure 3). These clinical findings are in agreement with embryological events, since obliteration of the first cleft begins in the ventral portion.

Figure 2. Membranous attachment (arrowheads) between the floor of the external auditory canal and the tympanic membrane. Left, Right ear. Right, Left ear.

Anomalies Strictly Located in the Parotid and External Auditory Canal Areas (n=18)

Anomalies Strictly Located in the Cervical Area (n=8)

Anomalies Located in Both Sites (n=13)

Figure 3. Triangle area where first branchial anomalies were located.

<table>
<thead>
<tr>
<th>Data</th>
<th>No. (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical symptoms</td>
<td></td>
</tr>
<tr>
<td>Auricular</td>
<td>12 (24)</td>
</tr>
<tr>
<td>Parotid</td>
<td>18 (35)</td>
</tr>
<tr>
<td>Cervical</td>
<td>21 (41)</td>
</tr>
<tr>
<td>Anatomical presentations</td>
<td></td>
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<tr>
<td>Fistula</td>
<td>11 (28)</td>
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<tr>
<td>Sinus</td>
<td>20 (51)</td>
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<tr>
<td>Cyst</td>
<td>8 (21)</td>
</tr>
<tr>
<td>Tympanic membrane attachment</td>
<td>4 (10)</td>
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</tbody>
</table>

* Two symptoms were associated in 10 patients, and 3 symptoms were associated in 1 patient.

In practice, diagnosis of first branchial cleft cysts located in or near the parotid gland can be difficult without surgical exploration. Isolated cysts accounted for 23% of the branchial cleft anomalies in our series. In the series of Olsen and colleagues, 85% of the cysts were located within the parotid gland, and the preoperative diagnosis in almost all these cases was a parotid tumor. Branchial cleft anomalies are rarely associated with other facial malformations, and diagnosis is difficult. Several classifications have been proposed to assist clinical diagnosis. Work classified first branchial cleft anomalies into 2 groups based on clinical and histological features: type I anomalies present as a cystic mass and are purely ectodermal, while type II anomalies present as a cystic mass and are mixed...
Type II anomalies present as a cyst, sinus, or fistula or any combination thereof and are of ectodermal and mesodermal origin. This classification can be useful in 2 situations. First, in patients with no previous infection or surgical intervention, isolated cysts can be classified as type I (9 cases in this series). However, the possibility that an infected or surgically drained isolated cyst may have formed a sinus or fistulous tract cannot be ruled out (4 other cases in this series). Second, for histological purposes, Work’s criteria distinguish type I lesions, which contain squamous epithelium but no adnexal skin structures or cartilage (7 cases in this series), from type II lesions, which contain squamous epithelium with adnexal skin structures or cartilage (19 cases in this series). Similarly, using these criteria, Belenky and Medina\textsuperscript{6} and Aronsohn et al\textsuperscript{5} were unable to classify 44% and 36% of cases in their series, respectively.

Our experience suggests that analysis of clinical manifestations (cervical, parotid, and auricular) and the findings of careful physical examination focusing special attention on the external auditory canal are more helpful than anatomical or histological classifications in achieving early diagnosis and management of first branchial cleft lesions. In the present study, physical examination quickly allowed discovery of a fistula of the external auditory canal in 17 patients (44%), and an asymptomatic membranous attachment between the floor of the external auditory canal and the tympanic membrane was found in 4 patients (10%). This anomaly, which, to our knowledge, has never been previously reported in the English-language literature, is an important diagnostic feature. Radiographic studies can also be a useful adjunct to diagnosis. Magnetic resonance imaging allows assessment of the extent of the anomaly, especially in the parotid area, and high-resolution computed tomographic imaging shows its exact relationship with the external auditory canal and the middle ear.

Given the sometimes subtle presenting symptoms, a surgeon who is unaware of these lesions may be tempted to perform a limited resection. This attitude inevitably leads to recurrence, and almost 50% of the patients in our series underwent previous unsuccessful treatment. In the series of Aronsohn and colleagues,\textsuperscript{5} incision and drainage had been performed on 10 of 11 patients. In the series of Finn and colleagues,\textsuperscript{7} 50% of the patients had previous infection, with from 1 to 7 incision and drainage procedures. In the series of Ford and colleagues,\textsuperscript{2} the mean number of operations before permanent cure was 2.4 per patient. In addition to being ineffective, repeated incision and drainage can cause scarring that makes dissection of the facial nerve more difficult. An attempt at excision by following the tract through a small skin incision can lead to paralysis of the facial nerve, as was the case in 1 of the patients in our series before adequate treatment.

Permanent cure can only be achieved by complete surgical excision with wide exposure of the lesion. Parotidectomy and exposure of the facial nerve are required in most cases; eg, it was required in 92% of the cases in our series. The major risk is facial nerve injury, especially in patients who have had multiple infections or surgical procedures.\textsuperscript{8} Facial nerve damage occurred in 2 of 5 patients in the series of Ford et al\textsuperscript{2} and in 6 (15%) of the 39 patients in our series (it was temporary in 5 of the 6 patients). Based on embryology, it is reasonable to expect that the defect would be superficial to the facial nerve. However, surgical findings indicate that this relationship is highly variable. A review of 73 cases (a combination of those reported in the literature\textsuperscript{6} and those in our series) showed that the anomaly passed medial to the nerve in 21 cases (29%) and was split around the nerve in 6 (8%). As previously reported,\textsuperscript{4,6} the isolated cysts in our series were always located above the main trunk of the facial nerve. Special care is necessary at the upper end of a fistula to remove not only the portion of the tract between the skin and cartilage of the external auditory canal, but also the flange of cartilage through which the tract passes (Figure 5).
CONCLUSIONS

First branchial cleft anomalies are uncommon, and recognition can be difficult. They should be suspected in patients with otorrhea, in the absence of chronic otitis, and an orifice communicating with the external auditory canal; in patients with a cutaneous opening located in the area of the neck defined by the hyoid bone below, the sternocleidomastoid muscle in the back, and the edge of the mandible in the front; and in patients with isolated cysts in the parotid gland that increase in size during an inflammatory period.

Early diagnosis and treatment are needed to avoid recurrent infection and secondary development of fistulous tracts. Permanent cure of first branchial cleft anomalies requires complete surgical excision, which usually necessitates identification and protection of the facial nerve. To ensure complete resection in the region of the external auditory canal, an area of skin and cartilage surrounding the fistula must be removed.

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REFERENCES