PHOTO ESSAY

An Atlas of Lumps and Bumps: Part 18

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Branchial Cleft Anomalies

At approximately the fourth week of intrauterine life, 6 pairs of branchial arches develop, of which 2 pairs are rudimentary.[1] The arches are mesodermal condensations on the lateral cervical area of the embryo.[2] These arches are separated by 5 invaginations of ectoderm on the outside (branchial clefts) and endoderm on the inside (pharyngeal pouches).[2] The second, third, and fourth clefts combine to form the cervical sinus of His. During the sixth embryonic week, the second branchial arch starts to grow caudally and eventually overgrows the third and fourth branchial arches by merging with the epipericardial ridge of the lower neck. As the branchial arches coalesce, the cervical sinus of His is obliterated.[2] Persistence or incomplete obliteration of the cervical sinus of His accounts for the presence of a branchial cleft anomaly.[2,3]

Branchial cleft anomalies account for approximately 40% of congenital neck masses and are the second most common cause of congenital neck masses in children, preceded only by thyroglossal cysts which account for approximately 43% of cases.[4] Gender distribution is similar for both male and female fetuses.[3,5,6] There is no racial predilection.[5,7] Branchial cleft anomalies occur more frequently in patients with a positive family history.[7] The occurrence is usually sporadic. Branchio-oculo-facial syndrome and branchio-oto-renal syndrome (also known as Melnick-Fraser syndrome) should be suspected if there are associated preauricular pits and multiple branchial cleft anomalies, especially if the branchial cleft anomalies are bilateral.[5,8,9]

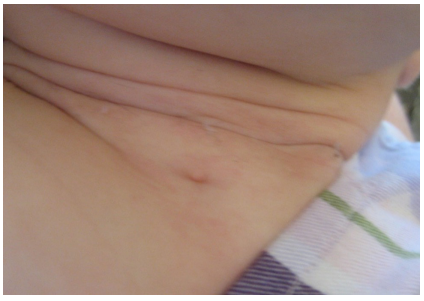

Figure 1. Branchial cleft anomalies may present as a sinus.

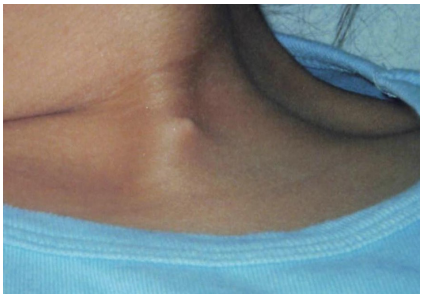

Figure 2. Branchial cleft anomalies may present as a cartilaginous remnant.

Approximately 95% of branchial cleft anomalies are caused by malformation of the second branchial cleft.[1,10] Branchial cleft anomalies may present as a sinus **(Figure 1)**, cyst, fistula, or cartilaginous remnant **(Figure 2)**, alone or in combination, in the lateral aspect of the neck anterior to the sternocleidomastoid muscle.[2,3,7,10,11] In one study, the most common presentation was a discharging sinus (59.25%) followed by a cystic neck swelling (33.3%).[12] A branchial cleft cyst typically presents as a nontender, mobile, and fluctuant mass located along the anterior border of the sternocleidomastoid muscle, usually just above the clavicle, although initially it may present as a visible punctum.[2,5,6,13] A branchial cleft sinus tract may communicate either externally with the skin as a visible punctum/opening or internally with the larynx or pharynx.[5] A branchial cleft fistula communicates externally with the skin as a visible punctum/opening and internally with the

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EDITOR'S NOTE:
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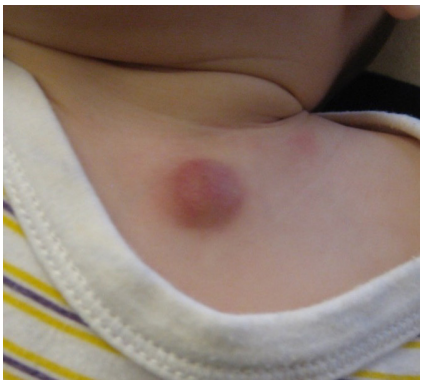

Figure 3. Branchial cleft anomalies may result in abscess formation.

larynx or pharynx.[5] The cutaneous punctum/opening may elevate on swallowing, forming a cutaneous depression similar to a dimple. This is referred to as a "dimple swallow" sign.[14] There may also be purulent discharge from the cutaneous punctum/opening.[5] A cartilaginous remnant usually presents as a stiff, elastic skin tag or mass that has a hyaline cartilage core. Approximately 98% of branchial cleft anomalies are unilateral, and of these, 83% to 97% are on the left side presumably consequent to asymmetrical vascular development.[15]

Although branchial cleft anomalies are congenital anomalies and are present at birth, they may not be obvious or symptomatic until later.[5] Some are detected when they become more prominent in late childhood. Other cases become apparent during intercurrent upper respiratory tract infections or when the cyst becomes infected. Generally, fistulae, followed by sinuses, tend to present at a younger age.10

The diagnosis of branchial cleft anomalies depends on good history taking, a focused examination, a high index of suspicion, and proper image studies. Ultrasonography, computed tomography, magnetic resonance imaging, and at times, sinography/fistulography may be used to confirm the diagnosis and the extent of involvement.[14]

Secondary bacterial infection occurs in approximately 10% of cases and may result in abscess formation **(Figure 3)**.[3,16] Occasionally, cystic compression of the upper airway may result in stridor and dyspnea.[5] The reported recurrence rate after surgical excision ranges from 3% to 7%.[3] Squamous cell carcinoma is a rare complication reported in adulthood.[2]

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