Branchial Anomalies: Etiology, Clinical Features and Management

Frederick K. Kozak & Juan Camilo Ospina

The pathologic conditions secondary to the branchial apparatus must be recognized and appropriately treated by the otolaryngologist. An understanding of the embryology related to this area of the head and neck is an essential component for the physician caring for these patients. The following is a short review of the clinical features and management of these conditions.

Microtia and Congenital Aural Atresia

Microtia is the most common major malformation of the pinna. Interuption of the normal formation of the hillocks of His results in microtia. The incidence is estimated to be between 1 in 1,000 and 1 in 8,000. The male to female ratio is 2.5:1. The majority of cases are unilateral and there are 4 types of microtia. **Type 1**: the external ear is slightly smaller than normal with minor subtle changes of the external pinna involving the helix and antihelix. **Type 2**: the ear is smaller and more abnormal than type 1 and all major structures are present but not fully. **Type 3**: the external ear is virtually absent with a small portion of cartilage present and frequently the lobule remains, but is anteriorly positioned. **Type 4**: anotia or absent external ear. Reconstruction relies on autologous rib grafts in a staged manner or the utilization of a prosthetic ear.

The external auditory canal develops from the first branchial groove. A failure of resorbtion and canalization results in the majority of cases of atresia/stenosis. The most complete classification regarding congenital aural atresia is that described by Schucneckt that details the severity of the involved structures as outlined below.

A: Mild meatal stenosis with canal cholesteatoma. Occassional pinna deformity. Normal or mildly abnormal middle ear structures.

B: More extensive stenosis, occassionaol microtia, moderate abnormality of middle ear structures.

C: Total aural atresia. Frequent microtia, bony atretic plate, relatively normal middle ear structures, moderate facial nerve abnormality

D: Total aural atresia, frequent microtia, total atretic bony plate, severly altered middle ear, frank facial nerve abnormality

The De la Cruz classification divides the abnormalities into major and minor malformations related to mastoid pneumatization, facial nerve position, presence of the oval window/footplate and status of the inner ear.

Candidacy for surgical correction and potential outcome revolves around the

Jahrsdoerfer grading system. A score of 8 points or greater predicts the best chance of success. Ten points are available with 2 for the presence of the stapes and 1 point for each of the following:oval window,middle ear space,facial nerve normal,presence of the malleus –incus, pneumatization of the mastoid, incus-stapes unit, round window normal, external ear appearance.

Preauricular Sinuses and Appendages

Preauricular sinuses and appendages (tags) constitute the most frequent minor deformity of the external ear. The prevalence of these is 0.8% and 0.2% respectively. Skin tags are thought to result from the abnormal fusion of the hillocks of His. They are generally localized in the pretragal region, but may extend out over the mandible. They may be multiple or single. Preauricular sinuses are keratinized epitheial tracts that end blindly or in a pseudocystic dilation without internal communication secondary to abnormal fusion of the Hillocks as well. Frequently there may be multiple fingers like extensions stemming from the initial tract. These may become infected via the accumulation of epithelial debris and blockage of the tract. In this instance, oral antibiotics may be effective in controlling the infection or if abscess formation occurs incision and drainage is indicated. Once infection takes place, complete surgical resection is indicated after the acute phase has subsided for a minimum of six weeks. When the tract is identified and closely adherent to the associated cartilage a small portion of cartilage should also be removed. In the majority of cases skin tags and preauricular sinuses occur in isolation but one must also consider the possibility of a number of associated syndromes such as the Branchio-Oto-Renal syndrome.

Branchial Cleft Anomalies

First Branchial Cleft Anomalies

Work's classification determines whether the anomaly is type 1 or type 2. **Type 1** is a membranous duplication of the external auditory canal of ectodermal origin. It is lined by keratinized squamous epithelium. It may present as a sinus, fistula, or cyst. A sinus is a tract lined by epithelium with one communication either externally to the skin or internally to mucosa. A fistula has both internal and external communication with the skin and mucosa. A cyst has no external or internal communication and is epithelial lined. The facial nerve is usually medial to the Type 1 defect. These may end blindly along the canal, into the canal or into the middle ear which is rare. **Type 2** anomalies have a membranous and cartilaginous duplication of the external auditory canal and are of ectoderm and mesoderm origin. They may present anywhere along the mandible and end either inferiorly or directly communicating with the external auditory canal. The relationship to the facial nerve is variable, but frequently the nerve is medial.

Imaging of this lesion can be helpful in surgical planning. Complete surgical excision is curative. The decision to operate may depend on whether or not the lesion has been infected previously. If infection has not occurred, similar to that seen with preauricular sinuses, some surgeons may consider a watchful approach. However, because of the high incidence of infection, early removal is usually recommended. Discussion with the patient/family regarding increased difficulty in removal post infection and risk to the facial nerve are imperative.

Second Branchial Cleft Anomalies

These are the most frequent of the branchial cleft anomalies. They may present as cysts, sinuses or fistulas. They are localized anterior to the STCM muscle in the upper third of the neck where they usually present as a cyst. When a sinus tract is present it can be in association with a cyst or simply a sinus tract alone. The length of the sinus tract is variable. As a fistula, the course of the tract is from the external opening on the neck, penetrating the platysma and then running parallel and lateral to the common carotid and the X cranial nerve. At the level of the carotid bifurcation the tract medializes after passing superiorly to the XII and IX cranial nerves and piercing the pharyngeal middle constrictor muscle with termination in the tonsillar fossa.

Third Branchial Cleft Anomalies

These present as either variable length sinuses or as fistulas. The course of this lesion commences at the skin in the inferior third of the neck, anterior to the SCN muscle and ascends through the playsma running parallel and lateral to the common carotid artery and the X cranial nerve. It then loops behind the internal carotid, loops back over the XII cranial nerve but inferior to the IX cranial nerve. It then continues its descent medial to the external carotid and perforates the thyro-hyoid membrane superiorly to the superior laryngeal nerve and into the piriform sinus.

Fourth Branchial Cleft Anomalies

These are extrememly rare with the tract (sinus or fistula) running a similar course to a third branchial cleft anomaly, however once descending it continues along the pathway of the recurrent laryngeal nerve and then looping anteriorly to posteriorly under the aortic arch on the left side or the subclavian artery on the right and then traversing back up along the route of the recurrent laryngeal nerve entering the larynx with the nerve or piercing the thyro-hyoid membrane into the piriform fossa.

Management of 2nd, 3rd and 4th Branchial Cleft Anomalies

Management may include imaging to determine the length of the sinus/fistula with a CT scan. Injection of contrast into the sinus at the time of imaging may assist in determining the length. Ideally, removal is undertaken prior to the first infection. Care regarding the associated nearby neuro-vascular structures as indicated above are important. Stepladder incisions to follow a longer tract can be helpful. Methylene blue injection at time of surgery may assist in the delineation of the tract to ease surgical removal. However, if the tract is interrupted during removal, spillage of the dye can then hamper the disection.

Pharyngeald Pouch Anomalies

First Pharyngeal pouch Anomalies

This entity has not been reported in humans.

Second Pharyngeal Pouch Anomalies

The derivation of the tonsillar fossa is from the second pharyngeal pouch. In the event of a persistent sinus affecting the tonsillar fossa the patient may present with recurrent unilateral tonsillitis. If found post tonsillectomy it is recommended that the sinus tract be cauterized if not too long and with the proviso that the nearby vascular structures are taken into consideration.

Third and 4th Pharyngeal Pouches

The thymus, thyroid gland, parathyroid glands and their associated vascular components are derived from these pouches. Patients with 22q deletion (Velocardial Facial Syndrome) may present with absence or abnormal functioning of these organs and the associated clinical picture. Third and fourth branchial pouch sinuses are rare. They may present as a lateral neck mass, recurrent acute suppurative thyroiditis or recurrent neck or thyroid abscesses. Fourth pharyngeal anomalies invariably are on the left side. Imaging with CT, MRI and barium swallow to determine the length of the associated piriform sinus fistula will assist in delineation of the pathology. Management also includes assessment of the length of the sinus via endoscopy and if short, consideration to cauterization can be undertaken. Hemithyroidectomy when indicated and neck exploration in order to remove the entire tract with complete surgical excision of the anomaly is usually curative. Utilization of methylene blue or cannulation with a fogerty catheter for tract identification has been described.

Thyroglossal Duct Cysts (TGDC)

The thyroid gland originates from the 4th pharyngeal pouch from the floor of the primitive pharynx caudal to the tuberculum impar at the base of the tongue. Between the 7th and 8th week of embryonic development the thyroid tuberculum descends to reach its definitive location anterior to the upper trachea. Communication between this tract and the base of the tongue is termed the thyroglossal duct. Normally this tract should disappear during embryogenesis. In the event of persistence a respiratory lined mucosal tract with a cystic mass will be present usually in the midline of the neck between the thyroid gland and base of the tongue. Occasionally the lesion may be off of the midline. On occasion they can be mistaken for a dermoid cyst or midline lymph node.

Imaging with ultra sound or CT will assist in delineating the lesion and establishing whether or not normal thyroid tissue is in the usual place. The majority of surgeons would recommend removal of a TGDC once the diagnosis is made. Removal prior to infection is much easier. If presentation is as an acute infection, it may settle with antibiotics. If presenting as an abscess, incision and drainage is required. Complete surgical removal after a minimum wait of 6 weeks for those presenting with either an acute infection or abscess is then recommended. The Sistrunk procedure is the most accepted approach to removal.

Recomended readings

1. Triglia J-M, Nicollas R, Ducroz V, Koltai PJ, Garabedian E-N. First Branchial Cleft Anomalies. Arch Otolaryngol Head Neck Surg. 1998;124:291-295
2. Pereira KD, Losh GG, Oliver, D, Poole MD. Management of anomalies of the third and fourth branchial puches. IJPO 2004;68:43-50.
3. Jahrsdoerfer RA. Congenital atresia of the ear. Laryngoscope 88 (suppl 13): 1-48, 1978. Gorlin RJ, Toriello HV, Cohen MM. Hereditary hearing loss and its syndromes Oxford Monographs on medical genetics No. 28. 1995.

4. Schuchtnecht HC. Congenital aural atresia. Laryngoscope 99:908, 1989.
5. Yeakley J, Jahrsdoerfer RA, Aguilar EA, et al. Grading system for the selection of patients with congenital aural atresia. Am J Otl 13:6,1992.
6. Work WD. Newer concepts of first branchial cleft defects. Laryngoscope 82:1581, 1972.
7. Liston SL. Fourth branchial fistula. Head Neck Surg 89:520-522, 1981.
8. Takai SI, Miyauchi A, Matsuzuka F, Kuma K, Kosaki G. Internal fistula as a route of infection in a acute suppurative thyroiditis. Lancet 4:751-752, 1979.
9. Feldman JI, Kearns DB, Pransky SM, Seid AB. Catheterization of branchial sinus tracts. Int J Pediatr Otorhinolaryngol 20:1-5, 1990
10. De la Cruz A, Linthicum FH Jr., Luxford WM. Congential atresia of the external auditory canal. Laryngoscope 95:421-427, 1985