Chapter 20: Branchial cleft anomalies, thyroglossal cysts and fistulae

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Branchial cleft anomalies and thyroglossal cysts and fistulae are the end result of defects in development in the neck area of the embryo. In this chapter an attempt is made to show how such defects occur and the mechanisms by which various well recognized clinical conditions are created. It should be remembered that the development of the neck is complex and that our knowledge of the rapidly changing anatomical relationships between the various structures is incomplete. Nonetheless, when embryological knowledge is combined with a study of established clinical conditions, some degree of certainty and logic can emerge.

Branchial cleft anomalies

Embryology

In the early embryo, the foregut develops between the brain above and the primitive heart below (Hamilton, Boyd and Mossman, 1972). The mouth is separated from the pharynx by the buccopharyngeal membrane which disappears around the end of the third week when a series of bars appears in the walls of the pharynx. These bars are formed by mesodermal condensations and are known as the branchial arches. The arches fuse ventrally, thus forming U-shaped structures which support the pharynx. Initially there are six of the arches, but the fifth is vestigial and rapidly disappears.

Four branchial pouches internally, and four branchial grooves (clefts) externally, separate the remaining five branchial arches. Each branchial pouch is lined by endoderm and each branchial groove by ectoderm, and these pouches and grooves are separated by a thin layer of mesoderm. In fish this endoderm-mesoderm-ectoderm layer breaks down so that a branchial cleft or gill-slit is formed. This does not normally occur in man but, should it do so, a branchial fistula may result.

A central core of cartilage develops in each arch and muscles differentiate from the surrounding mesoderm. Each arch is supplied by a cranial nerve and by an artery (aortic arch artery) which connects the ventral aortic sac to the dorsal aorta. Our knowledge of the exact derivatives of the arches is detailed in the more cranial arches, but becomes progressively less so in the caudal arches.

The first branchial arch gives rise to the maxilla, incus, malleus, anterior ligament of malleus, sphenomandibular ligament and mandible. The muscles are the muscles of mastication and the nerve is the mandibular branch of the trigeminal nerve. The artery is the first aortic arch artery from which is formed the maxillary artery.

The second arch forms the stapes, styloid process, stylohyoid ligament and the lesser cornu and upper part of the body of the hyoid bone. Its muscles are the muscles of facial expression which are supplied by the facial nerve. The artery is the second aortic arch artery which, rarely, may persist as the stapedial artery.
The third arch gives rise to the greater cornu and lower part of the body of the hyoid bone. Its nerve is the glossopharyngeal nerve and its artery is the third aortic arch artery which persists as part of the internal carotid artery.

The fourth and sixth arches form the laryngeal cartilages and the muscles of the pharynx and larynx which are supplied by the superior laryngeal (fourth arch) and recurrent laryngeal (sixth arch) branches of the vagus nerve. The fourth aortic arch artery forms the adult aorta on the left and the subclavian artery on the right while the sixth arch artery becomes the pulmonary trunk.

Between the branchial arches are the branchial pouches internally and the branchial grooves externally.

**First branchial pouch and groove**

The first pouch grows laterally to form the eustachian tube and middle ear while the groove is deepened to form the external auditory meatus. The pouch and groove meet at the tympanic membrane where they are separated by a thin layer of mesoderm. This mesoderm persists as the middle fibrous layer of the tympanic membrane. Note that only the dorsal part of the first groove takes part in forming the external auditory meatus. The remainder of the groove normally disappears but may persist as a preauricular sinus or a collaural fistula. The pinna is formed from a number of tubercles which appear at the dorsal ends of the first and second branchial arches. These tubercles surround the dorsal end of the first groove which is to form the external auditory meatus.

**Second, third and fourth branchial pouches and grooves**

The dorsal part of the second pouch contributes to the middle ear while the ventral part forms the supratonsillar fossa. The dorsal part of the third pouch forms the inferior parathyroid gland while the ventral part forms the thymic duct. The dorsal part of the fourth pouch forms the superior parathyroid gland while the ventral part probably contributes to the thyroid gland.Externally, the second branchial arch grows caudally and covers over the third, fourth and sixth arches, thus creating a deep pit or sinus lined by ectoderm. The opening to this cervical sinus is normally closed by fusion of its lips so that an ectoderm-lined cystic space is produced; later the cyst is resorbed and disappears.

**Pathogenesis and pathology**

Abnormal development of the branchial pouches and grooves may result in cysts, sinuses or fistulae.

A cyst is defined as a collection of fluid in an epithelium-lined sac. It may occur when part of a branchial groove or pouch becomes separated from the surface and fails to resorb. A cyst derived from a branchial groove will be lined by squamous epithelium; one derived from a branchial pouch will be lined by respiratory epithelium which may undergo squamous metaplasia after recurrent infections.
A sinus is a blind-ended track leading from an epithelial surface into deeper tissues. Such a sinus will occur when a branchial groove or pouch fails to resorb and remains open onto its epithelial surface.

A fistula is an abnormal communication between two epithelial surfaces. A branchial fistula is the human equivalent of the gill-slit in fishes and passes from the skin externally to the pharynx or larynx internally.

It is important to note that inclusion dermoids can closely mimic cysts derived from branchial grooves. In both cases the cyst will be lined by squamous epithelium and may contain skin adnexae. Only if cartilage is present can the cyst be certainly ascribed to a branchial groove origin. Inclusion dermoids occur especially in relation to the tubercles which appear at the cranial ends of the first and second arches and form the pinna. Here it is often impossible to be certain whether a cyst or sinus is an inclusion dermoid or a true branchial groove abnormality.

**Anomalies of the first branchial cleft**

Abnormal development may result in periauricular sinuses, cysts and collaural fistulae. Accessory tragi are also conveniently considered here although they develop from branchial arch tissue rather than the branchial cleft itself.

**Accessory tragi**

Accessory tragi are the result of anomalous growth of the tubercles of the first or second branchial arches and are therefore not true cleft anomalies. They are usually found in the preauricular region but may also occur anywhere along a line passing down to the sternoclavicular joint. They often contain cartilage and may be associated with other first and second arch abnormalities such as cleft palate and mandibular hypoplasia. If they do not contain cartilage they may be indistinguishable from a simple skin tag. Unsightly accessory tragi should be removed.

**Periauricular sinuses and cysts**

It may be difficult to decide whether these sinuses and cysts are inclusion dermoids resulting from epithelium trapped between the developing auricular tubercles or whether they are remnants of first branchial groove epithelium which has failed to resorb. Attempts have been made by Work (1972) and Batsakis (1980) to support one or other pathogenesis but the present author remains unconvinced, especially as there may be no pathological differences between the two. Perhaps one should simply state that superficial lesions may be a consequence of either, while the deeper lesions and especially those passing to the nasopharynx are more certainly branchial cleft in origin.

The sinus or cyst is usually preauricular in site, although inferior and posterior lesions do occur. The commonest lesion is the preauricular sinus with its opening just in front of the ascending limb of the helix. Preauricular cysts are less common and, unless large, may only present when they become infected. An infected cyst may rupture or be drained onto the surface and thus be converted into a sinus. Both sinuses and cysts are lined by squamous
epithelium and may contain skin adnexae in their walls. Many preauricular sinuses cause no trouble or may be kept quiescent by regular expression of any sebaceous material that collects. Others, however, become infected and cause recurrent pain, swelling and offensive discharge. Similarly, preauricular cysts often cause no symptoms and simply appear as an incidental finding in a routine otolaryngological examination. If symptoms are sufficiently troublesome, excision of the cyst or sinus with its track must be considered. The difficulty, of course, is that an apparently simple cyst or sinus may have extensive and deep branching ramifications which pass close to the facial nerve. The patient or parent, must fully understand the risk of facial nerve damage before agreeing to operation. He or she must be sure that the severity of symptoms justifies the risk of facial paralysis.

At operation, in the case of a cyst, a vertical incision is made over the cyst just anterior to the pinna. In the case of a sinus a similar incision is made but it should be split to include the opening of the sinus. Injection of the sinus with methylene blue may be of help, but care should be taken as any extravasation of dye outside the track will make dissection more difficult. The vertical incision can be extended into a standard parotidectomy incision if necessary. Dissection should now proceed medially until the whole cyst or sinus has been excised. Usually the track peters out above the level of the bony tympanic plate but, if it passes deeply into the parotid gland, it is best to carry out a superficial parotidectomy, displaying the facial nerve and its branches. It will now be possible to excise the track completely while preserving the facial nerve.

**Collaural fistula**

A collaural fistula is the least common of the first branchial cleft anomalies. It runs from the external auditory meatus or tragal notch down into the neck where it opens at a point between the angle of the mandible and the sternomastoid muscle. The fistula is caused by a failure of resorption of the ventral part of the first branchial groove. The fistulous track runs through the parotid gland and may pass medial to, lateral to, or through the facial nerve.

If the fistula is causing sufficient symptoms it should be excised. A modified parotidectomy incision is made which should include the ear opening at its upper end and the neck opening at its lower end. After the facial nerve has been displayed by a superficial parotidectomy, the track can be dissected and excised. It must be remembered, however, that recurrent infections and previous attempts at removal often cause extensive scarring and cystic masses. The facial nerve may be embedded in such tissue rendering it likely to damage. Patients *must* be warned of a possible facial paralysis.

Lastly, it should be noted that it is possible for the endoderm-mesoderm-ectoderm layer between the first branchial groove and first branchial pouch to disappear so that the two become continuous as a branchial fistula. Should such a fistula persist there will be an internal opening in the region of the eustachian tube orifice and the fistula will pass to the surface between the internal and external carotid arteries. The author has no personal experience of such a case, but suggests that the medial extent of the fistula could be explored in the same was as other lesions in the parapharyngeal space.
Anomalies of the second branchial cleft

Anomalous development of the second branchial cleft can result in fistulae, sinuses or cysts. A fistula occurs when the cervical sinus persists and the layer of endoderm-mesoderm-ectoderm between the second branchial pouch and groove breaks down. If the fistulous track is incomplete an internal or external sinus is formed. Rarely, a true branchial cyst is caused by incomplete resorption of the cervical sinus after closure of its lips. Most so-called 'branchial cysts', however, are probably due to epithelial inclusions in lymph nodes; this controversy, and 'branchial cysts' in general, are further discussed in Volume 5, Chapter 15.

A second branchial cleft fistula opens externally into the lower third of the neck just anterior to the sternomastoid muscle. This external opening will have been present since birth unless it has been produced by incision and drainage of an abscess. Other members of the patient's family may be affected and the lesion is occasionally bilateral. Recurrent infection with abscess formation may occur in the fistulous track or there may be an intermittent clear mucoid discharge through the external opening onto the skin of the neck. Pathologically, the fistula consists of a muscular tube lined by respiratory or squamous epithelium, the latter being more common after recurrent infection. There are often cystic dilatations along the course of the fistula and the submucosa may contain glandular elements as well as nerves and lymphoid tissue.

Fistulae which are the site of recurrent infection or discharge should be excised, but other fistulae can be left unless the external opening is cosmetically unacceptable. The extent of the track can be assessed preoperatively by injection of a radiopaque dye. A complete fistula is uncommon and most sinuses end well before the pharynx is reached. At operation, a horizontal skin crease incision is made to include the external opening. Dissection follows the track as it pierces the investing layer of deeper cervical fascia and ascends along the carotid sheath. After recurrent infection the track may be firmly adherent to the internal jugular vein or carotid artery so it is best to define these structures carefully as dissection proceeds. In long necks, and especially in complete fistulae, a further horizontal incision will be needed higher in the neck. The track is delivered into the upper incision and can be followed as it passes to the pharyngeal wall. The track must pass between the internal and external carotid arteries and it will be cranial or anterior to the glossopharyngeal and vagus nerves which are the nerves of the third and fourth arches. To ensure that symptoms will not recur, the whole track should be excised up to and including its opening into the pharynx.

Anomalies of the third and fourth branchial clefts

Third and fourth branchial cleft fistulae are rare and only a handful have been reported in the literature.

The author has treated one patient with a third cleft fistula and the case history will serve as a description.

The patient was a 12-year-old girl who had had 13 operations for drainage of recurrent neck abscesses. She presented to the author with a discharging opening in the neck just anterior to the sternomastoid muscle. At operation a fistulous track was found which passes between the common carotid artery and the vagus nerve and ended in the pyriform fossa. The
track was completely excised and histological examination showed it to be lined by stratified squamous epithelium.

A fistula of the fourth cleft would have to pass caudal to the arch of the aorta or right subclavian artery and end in the upper oesophagus or pyriform fossa. Isolated branchial cleft remnants have been described along this anatomical pathway (Downey and Ward, 1969; Tucker and Skolnick, 1973) but no complete fistula has yet been described.

**Thyroglossal cysts and fistulae**

*Embyology*

Towards the end of the third week of embryonic life a thickening of endoderm appears at the site of the tuberculum impar in the floor of the primitive pharynx. This endodermal thickening soon becomes evaginated to form the thyroglossal duct which descends into the neck between the first and second branchial arches so that it comes to lie in close relationship to the primitive aorta. Later the duct solidifies and is then known as the thyroglossal tract. When the thyroglossal tract reaches the front of the trachea it becomes bilobed to form the two thyroid lobes which are connected by the isthmus; each thyroid lobe may also receive a contribution from the ventral part of the fourth pharyngeal pouch. Normally, the rest of the tract disappears leaving the foramen caecum at the base of the tongue as the only adult indication of its place of origin.

Any part of the thyroglossal tract may persist into adult life. Much the commonest finding is a persistence of the lowest part of the tract as the pyramidal lobe of the thyroid gland. Less frequently, the tract may fail to descend into the neck from the base of the tongue so that it persists as a lingual thyroid. Because of the early relationship of the tract to the aortic arch, islands of thyroid tissue have also been found in the superior mediastinum. The most common clinical condition resulting from persisting tract remnants is the thyroglossal cyst. The precise embryology of the thyroglossal tract is fundamental to an understanding of surgical treatment and so will be described in detail.

As mentioned previously the tract descends into the neck between the first and second branchial arches. This means that it must descend between the developing mandible (first arch) cranially and the hyoid bone (second and third arches) caudally. The track passes into the neck, cranial or anterior to the hyoid bone and laryngeal cartilage.

Frazer in 1940 discussed the intimate relationship between the tract and the hyoid bone. He noted that the hyoid bone changes from an ovoid shape in the embryo to a crescentic shape in the adult. He suggested that this change of shape was a result of the downward pull of the strap muscles producing a downward projection of the body of the hyoid bone which thus indented the tract. In the adult, therefore, a persisting thyroglossal tract will pass down in front of the hyoid bone and then hook up around its inferior border to lie posterior to the bone before finally descending to the isthmus. Several authors (Ward, Hendrick and Chambers, 1949; Lawson and Fallin, 1969) have stated that the tract may descend posterior to or even through the hyoid bone. They do not, however, supply pathological evidence nor do they refute the embryological studies of Frazer (1940) and Hamilton, Boyd and Mossman (1972) which show conclusively that the tract descends cranial
or anterior to the second and third branchial arches in which the bone develops. Ellis and van Nostrand (1977) studied 30 embryos at varying stages of development, 200 adult larynges and 20 thyroglossal cyst specimens; in no instance did the tract pass down posterior to or through the hyoid bone. They suggested that reports of the tract passing through the hyoid bone could be due to misinterpretation of pathological specimens. All are agreed, however, that the tract is intimately related to the bone and that attempts to dissect the tract from its surface are likely to fail. The best way to remove all tract remnants and thus avoid recurrence is to excise the central part of the hyoid bone as recommended by Sistrunk in 1920 (see below).

Clinical features and management of thyroglossal tract remnants

Lingual thyroid

The thyroglossal tract may fail to descend into the neck so that the adult thyroid gland comes to lie at any point from the foramen caecum to the front of the trachea. The most common clinical condition is the so-called lingual thyroid. These patients usually present in childhood when a symptomless lump is noticed on the base of the tongue. Obstructive symptoms such as dysphagia or dysarthria are rare and usually occur only when the lump enlarges as a result of pregnancy, thyrotoxicosis or neoplasia. A thyroid scan must be performed if surgical excision is contemplated as the lingual thyroid may be the only functioning thyroid tissue present. Usually, however, no treatment is required and patients can be confidently reassured.

Thyroglossal cysts

Thyroglossal cysts occur equally in men and women and are usually noted in childhood, although they may present at any age. The cyst can occur at any point along the path of the tract from the base of the tongue to the thyroid isthmus, the commonest site being just above, or just below the hyoid bone. Most cysts are midline, but those at the level of the thyroid cartilage may be pushed to one or other side, usually the left.

Most patients present complaining of a symptomless lump in the midline of the neck. Examination will show that it is freely mobile from side to side and that it may transilluminate. The lump will rise on swallowing (on account of its attachment to the hyoid bone via the thyroglossal duct) and will also rise on protrusion of the tongue (because of its attachment to the base of the tongue via the thyroglossal tract). No other midline neck lump rises on tongue protrusion, so this physical sign is virtually pathognomonic. Some patients present with acute infection and abscess formation which may result in a sinus or fistula with intermittent discharge of a clear glairy mucus. Such a sinus or fistula is always acquired and may also be caused by inadequate surgical excision.

If the cyst is cosmetically unacceptable or is the site of recurrent infection and fistula formation it should be excised. Gross and Connerley (194) have argued that small symptomless cysts may be left in situ. However, many cysts do eventually become the site of recurrent inflammation and, very occasionally, a carcinoma may develop within the cyst. In most cases, therefore, surgical excision is the treatment of choice. A preoperative thyroid scan should be performed and this will usually show a normal thyroid gland in the normal
position. Perhaps surprisingly, it is uncommon to find functioning thyroid tissue in relation to the cyst.

**Sistrunk's operation for the removal of thyroglossal cysts**

In 1920, Sistrunk described his technique for the removal of thyroglossal cysts. He had found that simple excision of the cyst was often followed by further cyst formation or a chronically discharging sinus at the operative site; he argued that these recurrences could be avoided only by removing the whole of the thyroglossal tract. To do this he advised excision of the central part of the body of the hyoid bone and a core of tongue muscle up to the foramen caecum. Using this technique on 270 patients at the Mayo Clinic, Brown and Judd (1961) were able to reduce the previously high recurrence rate to only 4%.

A horizontal skin crease incision is made at the level of the cyst including, if necessary, the external opening of any sinus or fistula. The cyst is easily defined lying anterior to the larynx and it will usually be possible to find a well-defined fibrous cord passing to the body of the hyoid bone. Using bone-cutting forceps, the body of the hyoid bone is transected on either side of the midline, thus freeing a central portion of about 1-2 cm in length. Patients seem to suffer no disadvantage through losing most of the body of the hyoid bone and there is no need to attempt to suture the cut ends of bone together. It may be possible to follow the fibrous cord superiorly through the tongue musculature to the base of the tongue but, more usually, the cord peters out above the hyoid bone. In all cases, however, a core of tongue tissue comprising parts of geniohyoid and genioglossus should be excised, thus ensuring that all tract remnants are removed. A useful manoeuvre at this stage is to place the index finger of the left hand in the mouth on the foramen caecum. This removes any uncertainty concerning which part of the tongue should be excised and allows dissection right up to the foramen caecum. Ideally dissection should stop just short of the mucosa at the foramen caecum. If the mucosa should be breached, a catgut purse string suture will produce satisfactory closure. During closure, great care should be taken to halt any bleeding and to drain the wound. A postoperative haematoma may cause respiratory obstruction and be life threatening. Close supervision is essential in the early postoperative period and stitch scissors and sinus forceps should be kept at the bedside so that any necessary drainage can be carried out.

Pathological examination of the cyst will show it to be lined by columnar epithelium, squamous epithelium or, occasionally, no epithelium at all. When the epithelium is columnar it is usually pseudostratified, ciliated and associated with mucous glands in the submucosa; the cyst itself contains mucus. When the epithelium is squamous it keratinizes so that the cyst contains keratin. After recurrent infections, epithelium is often absent and the cyst contains inflammatory exudate. Lymphoid tissue is not normally found in the cyst wall (contrast 'branchial cysts'), so that infection of the cyst is probably either blood-borne or spreads down a patent thyroglossal tract from the foramen caecum. The cyst may be in continuity with a tubular or solid thyroglossal tract and the tract may be duplicated or branching, emphasizing again the importance of excising a wide core of tongue tissue with the specimen.

Thyroid carcinomata are occasionally found in thyroglossal cysts. Page et al (1974) reviewed the literature and found 656 such cases, all of which were papillary carcinomata.
Most had presented as simple cysts and were diagnosed only on pathological examination. Treatment is adequate surgical excision followed by suppressive doses of thyroid.