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Embryology of congenital neck masses

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Knowledge of the embryogenesis of the head and neck is essential to the evaluation and management of congenital neck masses. In this article, we describe the embryology of the most common congenital neck masses: branchial apparatus and thyroglossal duct cysts. The anatomy of both branchial clefts and pouches is reviewed along with the histology and descent of the thyroid from its origin at the foramen cecum of the tongue. This article serves as the foundation for understanding the anatomy surrounding surgical resection of these lesions, which will be discussed in later articles.

Introduction

The branchial arches, also known as pharyngeal arches, consist of 5-6 pairs of fingerlike embryonic structures that develop into the face, neck, and pharynx (Figure 1).[1] They are composed of mesenchyme, which is derived from mesoderm, somites, and neural crest cells.[1] Branchial arches appear between the fourth and seventh week of gestation on each side of the pharyngeal foregut.[1] Between each arch is an indentation called a branchial cleft, which is composed of ectoderm. Lateral outpouchings of the foregut oppose the branchial clefts and are known as pharyngeal pouches. These are composed of endoderm. Each branchial derivative is numbered in cranio-caudal succession.

Aside from the fifth branchial arch, which undergoes resorption or never forms, the branchial arches give rise to the structures of the head and neck.[2,3] Each branchial arch receives cranial nerve innervation because of its proximity to the brainstem (Figure 2). The branchial arch couples with the adjacent cranial nerve and retains this innervation regardless of later migration in the developing head and neck. The specific bony, cartilaginous, vascular, and muscular derivatives of each branchial arch are outlined in Table 1. These are different from those of the endodermal pharyngeal pouches, which form glandular tissue and contribute to the upper aerodigestive tract. The derivatives of pharyngeal pouches are listed in Table 2.

Branchial cleft anomalies

Branchial cleft anomalies are the second most common congenital lesions of the head and neck and accounted for one-third of all congenital neck masses in one series.[1,4] They may present as cysts, sinus tracts, fistulae, or cartilaginous remnants.[1] They are bilateral in 2%-3% of cases with a familial component.[5] Although the cause of formation remains unclear, most believe that these anomalies result from remnants of the branchial apparatus, particularly the cleft.[6]

Because of the complex growth and migration pattern of the branchial arches, anomalies can present anywhere from the upper periauricular area to the clavicles (Figure 3).

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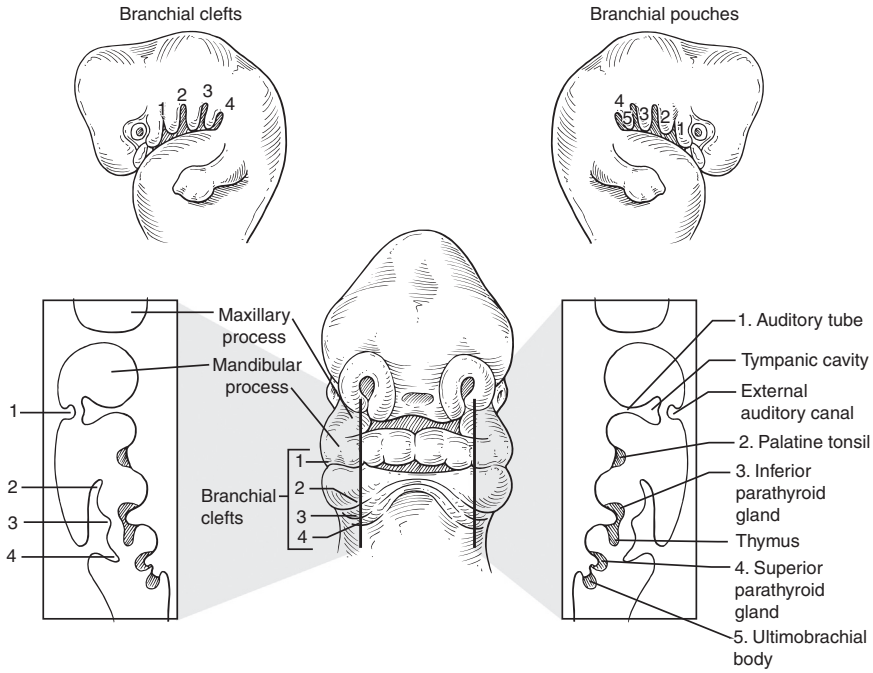

Figure 1 Diagram depicting branchial cleft and pouch derivatives.

Sinuses and fistualae often present as chronic draining lesions whereas cysts present as soft, compressible masses that may become inflamed or form an abscess during an upper respiratory tract infection.[7] In addition, many craniofacial syndromes can result from abnormalities in the branchial arches and pharyngeal pouches during embryonic development, including Treacher Collins syndrome, Goldenhar syndrome, branchio-oto-renal syndrome, Pierre Robin sequence, and DiGeorge sequence.

First branchial cleft anomalies

Anomalies of the first branchial cleft are rare, accounting for only 1%-4% of all branchial cleft anomalies.[8] They often include cysts or sinuses around the ear or near the angle of the mandible. However, they may be located anywhere along the tract, which begins in the submandibular triangle, rises to the parotid salivary gland, and ends in the external auditory canal.[7,9]

Because the first branchial cleft forms the external auditory canal, stenosis, atresia, and duplication of the canal may result. There are two types of first branchial cleft anomalies based on pathogenesis.[10]

(1) Type I anomalies consist of only ectodermal tissue. They involve duplication of the membranous ectodermal external auditory canal, which results in cystic masses adjacent to the canal.
(2) Type II anomalies are comprised of ectodermal and mesodermal portions of the canal. They may present as cysts, sinuses, or fistulae near the angle of the mandible.

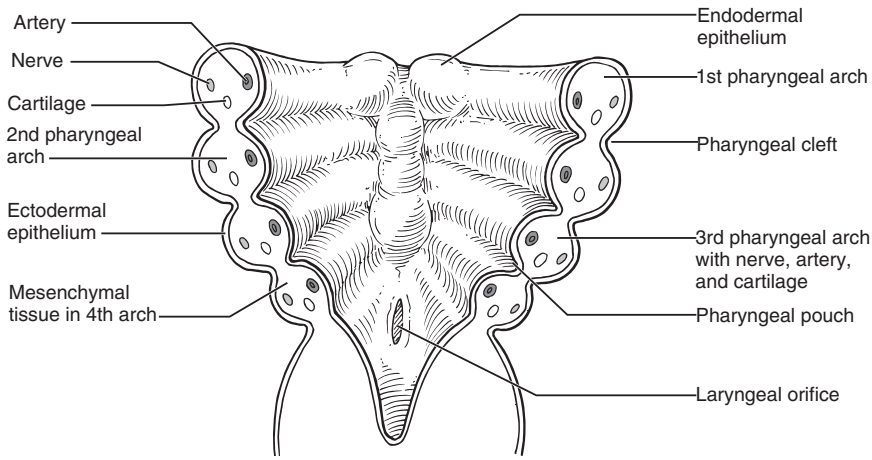

Figure 2 Branchial arches with their corresponding arteries, nerves, and cartilage embedded within mesenchyme. Note the location of ectoderm and endoderm germ layers and their relationships to branchial clefts and pouches, respectively.