Common Pediatric Head and Neck Congenital/ Developmental Anomalies

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KEYWORDS

• Congenital • Developmental • Neuroradiology • Pediatric • Genetic • Head & neck

KEY POINTS

- Identifying the location of pathologic entities relative to normal anatomic structures helps hone differential diagnosis and guides surgical approach.
- Clinical history, patient age, entity location, and the presence of associated anomalies all play important roles in refining a differential diagnosis.
- Many congenital pediatric head and neck disorders have pathognomonic imaging features which allow an interpreter to virtually make a diagnosis prior to biopsy.
- Although some lesions may seem aggressive, such as fibromatosis colli, they are considered to be do-not-touch lesions.
- A thorough understanding of pathologic entities is necessary to know when additional imaging studies are prudent.

INTRODUCTION

The complex nature of head and neck anatomy, in conjunction with the myriad of neonatal and early childhood head and neck disorders, often creates a diagnostic dilemma for primary pediatric care providers. Although adult head and neck disorders are primarily neoplasms such as squamous cell carcinoma, lymphoma, or melanoma, pediatric disorders are more often congenital/developmental anomalies such as infantile hemangioma, fibromatosis colli, thyroglossal duct cysts, or branchial apparatus anomalies. Clinicians are often able to determine with a high degree of certainty what a lesion may represent based on presentation and location. For instance, a warm, red or strawberrylike cutaneous discoloration most likely represents a benign infantile hemangioma, in which case watchful waiting or a simple ultrasonography (US) is all that may be needed. However, if a child presents with asymmetric cervical soft tissues, enlarging neck mass, or changes in feeding, a more rigorous diagnostic imaging work-up may be warranted.

When possible, most head and neck diagnostic evaluations in neonates, infants, and young children begin with US. This modality provides real-time information about blood flow direction and velocity, in addition to characteristic of the disorder and its relationship to adjacent structures. In addition, US is a cost-effective, readily available, and quick diagnostic imaging modality. In cases in which US does not completely characterize a lesion, or if the suspected entity is thought to

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originate within the deep cervical soft tissues, a computed tomography (CT) scan or MR imaging scan may be needed.

Although CT exposes patients to ionizing radiation, it is a more cost-effective and a faster imaging technique than MR imaging. In addition, it provides exquisite detail of involved osseous structures. MR imaging provides greater soft tissue evaluation and allows the determination of subtle soft tissue findings such as perineural tumor spread. However, patients may be subjected to conscious or full sedation, which have inherent risks. Radiologists and clinicians should work together to determine the most efficacious diagnostic work-up. This article reviews some of the commonly encountered pediatric head and neck anomalies, how to use a prudent diagnostic work-up, relevant anatomy, treatment options, and pearls that referring physicians should consider.

PATHOLOGY

Thyroglossal Duct Cyst

Thyroglossal duct cyst (TGDC) is a benign cystic mass occurring in the midline between the foramen cecum at the base of the tongue and the thyroid bed in the infrahyoid neck. It is the most common congenital neck malformation and is found in 5% to 10% of the population at autopsy. Presence represents failure of involution of the thyroglossal duct with persistent secretion via the epithelial lining. Most cases are spontaneous without a significant gender predilection. However, a rare autosomal dominant form, occurring most commonly in women, has a strong association with developmental thyroid anomalies.[1–3]

TGDC usually presents before 10 years of age as recurrent, intermittent swelling following recent respiratory infection. In rare cases, a lingual TGDC may lead to airway obstruction in the neonate. Differential considerations are broad and include additional cystic neck masses such as abscess, venolymphatic malformation, dermoid/epidermoid, or laryngocele.[4,5]

Physical examination shows a midline or paramedian palpable and compressible mass along the ventral neck. Approximately 25% occur in the suprahyoid neck, 25% in the infrahyoid neck, and nearly 50% at the hyoid bone. Treatment is via complete surgical resection. Location relative to the hyoid bone guides surgical approach. In most cases, a small section of the midline hyoid bone is resected to minimize recurrence.[6,7]

US is the imaging modality of choice for initial evaluation (see **Box 5**). A typical TGDC is midline along the ventral neck, well circumscribed, has anechoic internal echoes representing simple fluid, and thin walls. Its relationship to the hyoid bone should be noted (**Fig. 1**). If atypical features such as calcification, thick wall, isoechoic/hyperechoic internal echoes, or prominent adjacent vascularity are present, additional imaging with CT or MR should be pursued. Given the rare occurrence of associated thyroid anomalies, localization of normal thyroid tissue should be performed at the time of initial examination. If a normal thyroid gland is absent, nuclear scintigraphy aids in further evaluation. Please refer to **Table 1** and **Boxes 1** and **2** for additional diagnostic imaging findings.[2,3,5]

Branchial Apparatus Anomalies

Although the detailed embryology of the formation and development of the branchial (pharyngeal) apparatus is beyond the scope of this article, a basic understanding of the organization of the branchial arches, pouches, and clefts and their derivatives is helpful in understanding the pathogenesis of some important congenital head and neck masses.[8] Knowing the relationship of branchial remnants to normal anatomic structures is important in defining the anomaly and its point of origin,[9,10] which becomes clinically relevant in avoiding operative complications related to damaging adjacent structures during complete resection of these anomalies, which is the treatment of choice. The branchial anomaly and its associated tract typically are present inferior to the derivatives of its own arch and superior to the derivatives of the adjacent arch.[11,12] A simplified summary of branchial arch, pouch, and cleft derivatives are contained in **Table 2** and imaging recommendations of suspected lesions are shown in **Box 3**.

The most common branchial apparatus anomalies involve the first and second branchial clefts, whereas anomalies of the third and fourth apparatuses are rare. Approximately 95% of branchial cleft anomalies are related to the second branchial cleft, which also represents the second most commonly encountered congenital neck mass behind the thyroglossal duct cyst.[13] Between 1% and 4% of branchial anomalies can be attributed to the first branchial cleft. The first and second branchial cleft cysts are discussed here. Cervical thymic remnants, which are anomalies of derivatives of the third branchial apparatus, are discussed separately.

First branchial cleft cysts can present as masses within the region extending from the external auditory canal to the angle of the jaw, including the parotid gland.[2] Lesions associated with the external auditory canal may present auricular swellings, fistulas, or otorrhea, and may be