Evaluation of congenital masses of the neck in children: embryological, clinical and radiological review

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Learning objectives

- To emphasize the clinical features and radiological findings of congenital neck masses.

- To understand the importance of knowledge of embryology and anatomy of the cervical region when evaluating these lesions.

- To know the most appropriate imaging procedures to investigate pediatric patients.

Background

A neck mass is a common finding in pediatric patients and it may result from congenital anomalies, inflammatory and/or infectious processes, or less commonly neoplasms. Congenital lesions are one of the most frequent etiologies and include anomalies such as vascular malformations, thyroglossal duct cyst, branchial cleft anomalies, thymus anomalies and dermoid cysts.

Imaging is increasingly demanded for its investigation and the role of the radiologist is to help differentiating the nature of the mass, establishing or at least narrowing the diagnostic possibilities, and evaluate the anatomic extent for those lesions requiring surgical treatment.

Thorough understanding of the embryologic features and anatomy of the cervical region provides essential clues to help the radiologist in the evaluation of each lesion.

Findings and procedure details

THYROGLOSSAL DUCT CYST

Thyroglossal duct cyst is the most common congenital anomaly of the neck and the second most common neck mass in children after inflammatory lesions.

Embriology:

The thyroid gland originates in the 3rd week of fetal development from a median endodermal thickening in the floor of the primitive pharynx. The thyroid primordium develops at the level of the foramen cecum of the tongue and the developing gland
passes anterior to the hyoid bone, laryngeal cartilages, thyrohyoid membrane and the strap muscles, reaching the expected midanterior neck location by the 7th week of gestation. The duct normally involutes by the 8th week. Remnants of the duct, which occur in approximately 7% of the population, may lead to congenital anomalies, such as ectopic thyroid tissue or cyst formation anywhere along the course of thyroid gland migration (Fig.1).

**Clinical presentation:**

Thyroglossal duct cysts usually manifest as an asymptomatic mass during childhood and lie within the anterior triangle of the neck, at the midline or just off the midline position, most being located in the infrathyroid neck adjacent to the hyoid bone. On clinical examination the neck mass will typically move upward when the tongue is protruded. When infected it can be a painful mass, which is the typical presentation in the adult population.

**Imaging findings:**

US is an important imaging study in the preoperative evaluation of this condition to confirm the cystic nature of the palpable mass and to demonstrate the presence of a normal appearing thyroid gland in the lower neck.

On US they appear as a well-defined, thin-walled, homogeneous anechoic mass with posterior acoustic enhancement or uniformly echogenic pseudosolid appearance due to the proteinaceous content of the cyst secreted by the epithelial lining. The absence of signal on color Doppler confirms its cystic nature (Fig.2). A thick cyst wall, heterogeneous echopattern and inflammatory changes of surrounding fat planes indicate infectious complication of the cyst (Fig. 3A).

There must be a balance between optimal diagnostic studies that answer the clinical questions and the lowest possible dose exposure and invasiveness. Thus, further investigation with cross-sectional techniques should only be used in the evaluation of complicated cases or when diagnostic dilemma remains.

On CT, uncomplicated thyroglossal duct cysts are well-defined low attenuation masses and on MRI they have fluid signal intensity. Varying degrees of attenuation and signal intensity are usually seen because of the presence of internal proteinaceous contents and there might be minimal peripheral contrast enhancement. If there is superimposed infection, it often appears with peripheral contrast enhancement, septations and change in attenuation/signal intensity or edema in the adjacent soft tissues.

On preoperative imaging work-up of the thyroglossal duct cyst the following aspects need to be considered by the radiologist: less than 1% contain thyroid carcinomas within the
cyst (most present in adulthood), therefore, any solid component requires fine needle aspiration cytology (Fig. 3B); normal thyroid tissue has to be identified in the anterior neck before surgery to prevent postoperative hypothyroidism; the relationship of the cyst with the hyoid bone must be determined in order to help the surgeon to completely excise the lesion, reducing the chance of recurrence.

**BRANCHIAL CLEFT ANOMALIES**

Branchial anomalies are one of the most common congenital masses in children and over 90% arise from the second branchial apparatus, with a predominance of cysts.

**Embriology:**

The branchial apparatus appears around the 4th week of fetal life and consists of 4 pairs of well-defined branchial arches and 2 rudimentary arches that are separated externally by ectodermal-lined grooves or clefts and internally by endodermal-lined pouches.

Incomplete obliteration of a portion of the branchial apparatus, predominately the cleft, is postulated to lead to branchial cleft anomalies such as cysts, sinuses or fistulae.

Branchial cleft anomalies may be classified as first, second, third, or fourth arch anomalies according to the pouch or cleft of origin.

Third and fourth branchial anomalies are rare. The remainder of this review will focus primarily on branchial apparatus cysts derived from the first and second branchial cleft once they represent the majority of branchial cleft anomalies.

The mandible and the portion of the maxillary process are derivatives of the paired first branchial arches. The first arch also contributes to the development of the inner ear, while the external auditory canal, Eustachian tube, middle ear cavity and mastoid air cells are derivatives of the first branchial clefts and pouches.

The second arch forms the hyoid bone, as the second pouch forms the epithelium of the palatine tonsil and the supratonsilar fossae.

Knowing this, we can predict the origin of the majority of branchial apparatus cysts and becomes easier to evaluate images of these lesions.

**Clinical presentation:**

First branchial cleft cysts account for up to 8% of all branchial anomalies and the typical clinical presentation is a middle-age patient with painless fluctuating preauricular mass or sinus tract, with history of recurrent infections.
Over 90% of branchial anomalies arise from the second branchial apparatus, as mentioned earlier. They are typically recognized in patients 10-40 years of age and usually manifest as a non-tender, soft mass deep to the anterior border of the sternocleidomastoid muscle and are more common in children and young adults.

**Imaging findings:**

First branchial cleft cysts should be suspected when there is a cystic mass adjacent to the external auditory canal, superficial or deep within the parotid gland or adjacent to the pinnae extending into the anterior neck to the level of the angle of the mandible. In pediatric age group anomalies of the first branchial apparatus commonly present as sinus tract or fistula in the parotid region near the external auditory conduct (Fig. 4).

The location of second branchial cleft cyst is the key to diagnosis and most often are situated in the submandibular space, at the anteromedial border of the sternocleidomastoid muscle, lateral to the carotid space and posterior to the submandibular gland (Fig. 5).

Third branchial cysts that occur in the upper neck usually lie in the posterior compartment of the neck, posterior to the common or internal carotid artery; in the lower neck they can be in the anterior triangle.

Most fourth branchial cleft anomalies are sinus tracts which arise from the pyriform sinus, through the thyrohyoid membrane and descent into the mediastinum, sometimes presenting as a perithyroidal abscess (Fig. 6).

In any imaging modality, the cysts will appear as simple or complicated cysts, depending on the nature of the fluid within the cyst: simple fluid, proteinaceous fluid, hemorrhagic fluid or fluid complicated by superimposed infection.

On CT, the centre of the mass will have an attenuation value similar to water and if the cyst is infected, a thickened enhancing wall will be present.

MRI better depicts the deep-tissue extent of the cyst, which varies from hypo-to isointense on T1-weighted images and hyperintense on T2-weighted images. Occasionally, in cases of first branchial cleft cysts, there will be a definable tract extending to the external auditory conduct or connecting a bilobed cyst that is best evaluated with MRI.

**TERATOMAS AND DERMoid CYST**

**Embriology:**
Teratomas and dermoid cysts are developmental anomalies involving pluripotential embryonal cells. There are two plausible theories to explain the existence of these lesions: one suggests isolation of pluripotential cells during embryogenesis and subsequent disorganized growth of these cells; the second theory holds that germ layers may be buried in deeper tissues at points of failed fusion lines.

Dermoid cysts are composed of ectoderm and mesoderm and represent the most common form of teratoma, accounting 20% of head and neck dermoids. They are covered with skin and contain epidermal appendages such as hair follicles, sebaceous glands and sweat glands.

Teratomas contain all 3 germ layers and rarely occur at the neck.

Clinical presentation:

Dermoid cysts are typically diagnosed before age of 3 years and when situated on the neck, they usually appear as a midline mass at the submental region and less commonly it lies over the hyoid bone and can move with tongue protusion mimicking thyroglossal duct cysts. Infection of dermoid cysts is uncommon. The sebaceous secretions result in slow enlargement of these lesions.

Teratoma involving the neck is often diagnosed on prenatal US or MRI as a rapidly enlarging lateral or midline neck mass. They are usually clinically apparent at birth as large neck masses that may lead to dysphagia and respiratory distress in the neonate. The mortality is high if not treated.

Imaging findings:

On US, the dermoid cyst is usually well-defined and anechoic with posterior acoustic enhancement in a midline position of the neck. It may exhibit a pseudosolid appearance with homogeneous internal echoes due to the presence of cellular material within the cyst. Others may have mixed internal echoes because of its fat content and may show the presence of osseous-dental structures, seen as echogenic foci with posterior acoustic shadowing.

On CT, dermoids are known to variably enhance as well as demonstrate fat and dystrophic calcifications. Globules of fat floating within the lesion and fat and/or fluid levels may be present.

US features of teratomas include solid and cystic structures within a heterogeneous mass, and calcifications are seen more frequently than cartilage and bone formation.

It is important to determine the depth and the proximity to the hyoid to plan the resection. Both CT and MRI clearly define their anatomical location, extent and internal appearance. On CT, these lesions can have a variable presentation. They can present
as a hypoattenuating, thin-walled unilocular mass with heterogeneous contents. At MR imaging, teratomas are hypo- to isointense on T1-weighted images and hyperintense on T2-weighted images.

**VASCULAR CONGENITAL LESIONS**

Vascular lesions are divided into malformations and tumors. Based on the dominant haemodynamic flow pattern, vascular malformations can be classified into low-flow lesions (lymphatic, venous, capillary, or combined) and high-flow lesions (arteriovenous malformations and arteriovenous fistulas). Sometimes, a combination of flow patterns can be found constituting a mixed vascular malformation.

- **Lymphatic malformations**

Lymphatic malformations are low-flow vascular malformations and represent the first most common cystic mass in newborns. About 60% are associated with genetic disorders such as Turner syndrome, Noonan syndrome and trisomies.

**Embriology:**

The lymphatic system begins to develop at the end of the 5th week of gestation, and the early lymph capillaries join to form a network of lymphatic vessels and lymph sacs.

The majority of lymphatic malformations occur in the neck, presumably as a result from anomalies in the development of the cervical lymphatic system that arises as paired jugular lymphatic sacs sprouting from the primitive jugular venous plexus. These jugular lymphatic sacs eventually form communications with the venous system and become the terminal portions of the thoracic duct and the right lymphatic duct.

**Clinical presentation:**

The majority of cervical lymphatic malformations are diagnosis before the age of 2, with less than 10% occurring in adults. Males and females are equally affected. Most of these lesions occur posterior to the sternocleidomastoid muscle, in the posterior cervical triangle, as a painless spongy-like mass. Usually they are slow growing, however sudden enlargement can occur following infection or haemorrhage.

**Imaging findings:**

Lymphatic malformations are classified as macrocystic (>1 cm), microcystic (<1 cm) and mixed cystic forms (Fig. 9), being the macrocystic the most common type.
On US, lymphatic malformations manifest as a uni- or multilocular cystic mass, containing fluid or debris/hemorrhage, with echogenic septations of variable thickness (Fig. 7A). Echogenic portions representing clusters of abnormal lymphatic channels and fluid levels can be observed.

Microcystic lymphatic malformations appear predominantly hyperechoic because of the multiple small interfaces and typically are more infiltrative and can be confused with solid soft-tissue masses (Fig. 8A, 8B).

Prior to surgery, the extent and relationship with the neighboring structures should be assessed.

On CT, lymphatic malformations appear as uni- or multi-septated, hypoattenuating and infiltrative masses involving the posterior triangle of the neck (Fig. 8C). Haemorrhage or infection causes an increase in attenuation. These lesions frequently do not respect fascial planes. Although there is no internal enhancement, cyst walls and septations usually enhance after administration of contrast.

MRI is better than CT to show the relationship of the mass to the surrounding soft tissues. The mass will be of high signal on T2 weighted images and contain flow voids, representing feeding and draining vessels (Fig. 7C). On T1-weighted images will demonstrate low or intermediate signal intensity (Fig. 7B). Infection or hemorrhage can modify the observed signal intensity of the cavities.

- **Venous malformations**

Venous malformations are low-flow vascular malformations that consist in dysplastic venous channels, and approximately 15% occur in the head and neck.

**Clinical presentation:**

Most are soft, compressible lesions with bluish discolouration if superficial, and are usually diagnosed clinically. Soft-tissue lesions are most frequently facial in location, the buccal region being the most common site followed by the mandibular space, sublingual space, tongue and orbit.

**Imaging findings:**

On US, they appear heterogeneous, hypoechoic lesions with sinusoidal spaces, and color Doppler reveals slow blood flow. Phleboliths may be detected and are characteristic of this vascular malformation. On MRI, venous malformation shows a characteristic high signal on T2-weighted fat-saturated sequences. Phleboliths and osseous involvement may also be identified by CT.
• **Capillary and Capillary-Venous Malformations**

Capillary malformations are present at birth in 0.3% of children and most often are localized in the head and neck. They represent areas of congenital ectasia of thin-walled small-caliber vessels of the skin. MRI is not usually required because the diagnosis is made clinically. MRI findings may show skin thickening and increased subcutaneous thickness.

Capillary-venous malformations are combined low-flow malformations formed from dysplastic capillary vessels and enlarged postcapillary vascular spaces. Imaging findings may be similar to venous malformations. Dynamic contrast-enhanced MRI typically show early homogeneous enhancement, whereas only delayed enhancement is seen in venous malformations.

• **Arteriovenous Malformation and Arteriovenous Fistula**

High-flow vascular congenital malformations include arteriovenous fistula and arteriovenous malformation.

Arteriovenous malformation are already present at birth in the early quiescent stage but often clinically progress with pain, ulceration or bleeding later in life following trauma or infection. They demonstrate a nidus of small vessels between the supplying artery and draining vein, and manifest as a red, pulsatile, warm mass with a thrill and may lead to bone overgrowth, arterial steal phenomenon, and cutaneous ischemia. MRI findings include high-flow enlarged feeding arteries and draining veins, which appear as large flow voids on SE images or high-signal-intensity foci on GRE images, without a well-defined mass.

Congenital arteriovenous fistula consists of a macroscopic arteriovenous connection and usually occurs in the head and neck. MRI shows the arterial and venous components as large signal voids on SE images or high-signal-intensity foci on GRE images, without a well-defined mass.

**HAEMANGIOMAS**

Infantile haemangiomas are the most common vascular tumor and the most frequent head and neck tumor of infancy. The haemangioma result from hamartomatous proliferation of capillaries, usually present within the first month of life showing a high proliferation index in the first year of life and then typically slowly involute over 5 to 10 years. Congenital haemangiomas are already fully formed at birth, and unlike infantile haemangiomas they do not express the glucose transporter isoform 1. Although similar
in appearance, they behave quite differently and include two types: rapidly involuting congenital haemangioma (RICH) and noninvoluting congenital haemangioma (NICH).

**Clinical presentation:**

Hemangiomas that are on the surface of the skin are typically non-compressible, pulsatile, flat and bright red to purple in color, while deep hemangiomas may be blue, purple, or even normal skin color if they are located deep under the skin surface.

**Imaging findings:**

Routine imaging is not required unless the clinical diagnosis is in doubt, or to determine the relationship to vital anatomical structures.

On US, in the proliferative phase they frequently appear as a non-specific echogenic mass with low resistance, high-flow arteries and veins, most often in the parotid or buccal space (Fig. 10). The size and number of vessels decreases in the involutional phase. CT typically shows a strongly, homogeneous enhancing lobulated mass. The MRI appearance is usually slightly hypo-intense on T1, iso- to hyper-intense on T2 with multiple serpiginous flow foids. Its lobulated appearance with thin septa is characteristic.

**ECTOPIC CERVICAL THYMUS AND THYMIC CYST**

Ectopic cervical thymus (ECT) and thymic cysts are rare causes of pediatric neck masses, with most lesions detected as an incidental imaging finding.

**Embriology:**

The thymus migrate caudally and medially along with the parathyroid from the third pharyngeal pouch. The paired thymic primordia detach from the pharynx, complete their inferior migration along the thymopharyngeal ducts and fuse beneath the thyroid. After its development is complete the thymopharyngeal duct eventually disappears.

Abnormal thymic development can result in cystic lesions or ectopic tissue along the neck. Theories explaining these anomalies include hyperplasia of undescended or sequestered cervical thymic remnants and persistence of the thymopharyngeal duct.

Therefore, ECT and thymic cysts can be found anywhere along their embryologic tract of descent from the angle of the mandible to the superior mediastinum, adjacent to the carotid sheath (Fig. 11).

**Clinical presentation:**
ECT and thymic cyst most often manifest as an asymptomatic lateral neck mass. On rare occasions, the ectopic gland cyst may become infected or hemorrhagic, which causes an acute increase in its size and mass effect on adjacent structures. If the lesion is large enough it can lead to airway compression and respiratory distress.

**Imaging findings:**

The anatomic location gives an important clue to the diagnosis. They typically appear next to the carotid sheath, deep to the middle third of the sternocleidomastoid muscle and usually extends into the retropharyngeal space or mediastinum.

US enables clear differentiation between solid and cystic lesions and may suggest the diagnosis, but these lesions are more easily evaluated with use of the multiplanar capabilities of MR imaging.

In the presence of ECT, US demonstrated a homogeneous, hypoechoic with no through transmission mass, on its usually location (Fig. 12A). On MRI, ECT mimic the signal intensity of normal thymus on T1- and T2-weighted images, appear homogeneous with mild enhancement and do not invade adjacent structures (Fig. 12B, 12C, 12D).

On US the cyst appears as a well-defined, anechoic cystic lesion or show intrallesional mobile debris if the fluid is hemorrhagic or proteinaceous (Fig. 13A, 13B). On MRI, the cystic content usually has low to intermediate T1-weighted and high T2-weighted signal intensity (Fig. 13C).

**FIBROMATOSIS COLLI**

Fibromatosis colli, also called sternocleidomastoid tumor of infancy, is a rare, benign soft-tissue tumor present in the neonatal period and characterized by abnormal proliferation of fibroblasts. The exact etiology is still unknown, however, a history of birth trauma and genetic factors may play a role in its development.

**Clinical presentation:**

Affected patients clinically present with torticollis. On examination it appears as a painless mass in the sternocleidomastoid muscle. The clinical course is usually that of spontaneous resolution over a period of 4-8 months; rarely it may require surgical treatment.

**Imaging findings:**
The diagnosis of fibromatosis colli is best made with US. On US, the typically features are a fusiform thickening and shortening of the sternocleidomastoid muscle. It usually appears as a homogeneous isoechoic or hypoechoic mass within the sternocleidomastoid muscle (Fig. 14). The mass can be seen to move with the muscle on real time US.

A confirmatory CT or preferably MRI may be required in doubtful cases for further evaluation.

Images for this section:

**Fig. 1:** Typical course of thyroglossal duct remnants.
**Thyroglossal duct cyst. (A.)** Sagittal midline neck US shows an homogeneous hypoechoic well-defined lesion with posterior acoustic enhancement with a pseudo-solid appearance in a 7 year old boy presenting with a painless midline neck mass. **(B.)** Sagittal midline color Doppler US shows absence of flow within the lesion confirming its cystic nature, in the same patient.

Fig. 2

**Thyroglossal duct cyst complications. (A.)** Axial midline neck US shows at the hyoid level an anechoic lesion containing some hypoechoic debris, with a thickened well-defined outer wall, and hyperechoic surrounding fat planes (infected TGDC in a 2 year old girl with painful midline mass neck). **(B.)** Axial midline color Doppler US shows cystic lesion with solid vegetations within it, suspicious of malignant complication in a 15 year old adolescent.

Fig. 3
First branchial cyst and fistula. A. US at the level of the left parotid region shows an hypoechoic definable tract connecting the skin surface to a deep bilobed cyst (*). B. Axial contrast-enhanced CT scan shows a low attenuation lesion with minimal peripheral contrast enhancement located within the left parotid gland, adjacent to the external auditory canal.

Fig. 4

Second branchial cyst. (A.) Axial US image depicts an homogeneous hypoechoic lesion with posterior acoustic enhancement containing some slightly echogenic debris, located on the left side of the neck at the level of the left submandibular region. (B. and C.) Coronal and axial contrast-enhanced CT scan showing a cystic lesion with homogeneous low attenuation non-enhancing content and a well defined wall located in the left submandibular region. It displaces the sternocleidomastoid muscle slightly posteriorly, the carotid artery and internal jugular vein posteromedially and sumandibular gland anteriorly.

Fig. 5
**Fourth branchial cleft anomaly.** Axial (A.) and coronal reformation (B.) contrast-enhanced CT images at the level of thyroid gland show an heterogeneous allongated low attenuation lesion with thickened enhancing irregular wall along (arrow) the left side of the pharynx. The lesion is in close relation with the left lobe of the thyroid gland and extends upward to the level of the left pyriform sinus, compatible with infected fourth branchial cleft anomaly in a 3 year old boy.

*Fig. 6*

**Lymphatic malformation (macrocystic).** (A.) Axial neck US shows a compressible, multilocular cystic lesion with intervening thin septa in the left posterior triangle in a neonate with prenatal diagnosis of neck cystic lesion. (B. and C.) Axial T1W GRE with fat sat MR image (B.) and coronal T2 TSE MR image (C.) of the same patient showing a large left sided mass involving the posterior triangle of the neck extending from the auricular lobule to the supraclavicular region, exhibiting low signal intensity on T1W and high signal intensity on T2W images. Despite its large volume, there is no signs of airway compromise.

*Fig. 7*
**Microcystic lymphatic malformation.** (A.) Axial lateral neck US shows an heterogeneous, mixed solid and cystic mass in a young adult. (B.) On color Doppler US only a few scattered flow dots are seen. (C.) Axial contrast enhanced CT showing a hypodense, homogenous nonenhancing lesion (arrow) in the lower left carotid region.

**Fig. 8**

**Mixed lymphatic malformation.** (A. and B.) Axial and sagittal contrast enhanced CT images in a 2 day old neonate showing a huge right sided cervicothoracic mass, heterogeneous, with cystic components, that does not respect neck compartmental anatomy, extending into the thorax and compressing inferiorly the heart and mediastinal great vessels. There is compression and contralateral deviation of the airway. An endotraqueal tube was inserted during ex-utero intrapartum treatment (EXIT procedure). (C.) Coronal T2W MRI performed at the age of 2 months, after initial debulking surgery and previous to a second surgical intervention. Heterogeneous cervicothoracic mass with solid and varying sized cystic components, still compressing and invading cervical and mediastinal vital structures but with less airway compromise than in initial CT.

**Fig. 9**
Haemangioma. (A.) US axial imaging of the left parotid region shows an intraparotideal hypoechoic heterogeneous well defined nodular mass with some cystic spaces. (B.) Color Doppler US shows the lesion to be markedly vascularized.

Fig. 10
**Fig. 11:** Diagram shows the course of the thymopharyngeal duct.
**Ectopic cervical thymus.** (A.) Sagittal US at the level of the suprasternal notch shows a homogeneous, hypoechoic solid mass. (B., C. and D.) Axial (B.), coronal (C.) and sagittal (D.) T2W MRI show a homogeneous hyperintense elongated structure (arrows) with similar signal intensity and contiguous to the intrathoracic left lobe of the thymus gland (*). This is the so-called cervical extension of the thymus.

Fig. 12

**Thymic cyst.** Right-sided neck axial (A.) and sagital (B.) US showing a multiloculated cystic lesion with thin walls and homogeneous hypoechoic fluid content in close relationship with the carotid sheath. (C.) Coronal T2W MRI revealing a large heterogeneous mass with multiloculated cystic areas in the right side of the neck, in close relationship with the carotid sheath, extending inferiorly into the superior mediastinum and connecting with the thymus.

Fig. 13
Fibromatosis coli. Longitudinal US image of a slightly heterogeneous isoechoic mass within the sternocleidomastoid muscle in a neonate.

Fig. 14
Conclusion

The differential diagnosis of a pediatric neck mass includes a wide range of pathologies. Knowledge of embryology, anatomy and characteristic imaging appearances of congenital masses is essential to easily recognize them.

After clinical examination, US is often the initial and first-line investigation since it is readily available and does not involve ionizing radiation. CT and MRI are usually required for further characterization and complete anatomic extent evaluation regarding surgical planning. MRI is usually preferred over CT due to its lack of ionizing radiation which is a major concern in children. Nevertheless, CT has a crucial role in acute conditions, such as complicated congenital cysts or large masses compressing the airways that may warrant urgent surgical intervention.

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