International Journal of Research in Medical Sciences
Rasool Z et al. Int J Res Med Sci. 2013 Nov;1(4):329-332
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: 10.5455/2320-6012.ijrms20131103

Review Article

Congenital neck masses: embryological and anatomical perspectives

Zahida Rasool[1]*, Ashfaq U. Hassan[2]

[1]Medical Consultant Islamic University of Science and Technology, Awantipora, Jammu and Kashmir, India
[2]Department of Anatomy, SKIMS Medical College, Bemina, Jammu and Kashmir, India

Received: 21 June 2013
Accepted: 4 August 2013

***Correspondence:**
Dr. Zahida Rasool,
E-mail: zahidarasool12@rediffmail.com

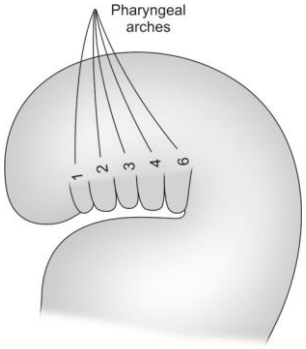
ABSTRACT

Neck masses are a common problem in paediatric age group. They tend to occur frequently and pose a diagnostic dilemma to the ENT surgeons. Although the midline and lateral neck masses differ considerably in their texture and presentation but the embryological perspective of these masses is not mostly understood along with the fundamental anatomical knowledge. The article tries to correlate the embryological, anatomical and clinical perspectives for the same.

Keywords: Torticollis, Cyst, Branchial, Hygroma, Sternomastoid, Pouch, Arch, Cleft, Branchial

INTRODUCTION

The common benign anomalies of the cervical region include cysts and sinuses, thyroglossal cysts, cystic hygromas, dermoid cysts, hemangiomas.[1,2] Nonlymphoid masses (cervical rib, thyroglossal cyst, branchial sinus or cyst, cystic hygroma, goiter, sternomastoid muscle tumor, neurofibroma) occur frequently in the cervical or neck region.



Cysts and sinuses in the neck may be formed along the course of the first, second, third, or fourth branchial clefts as a result of improper closure during embryonic life. Second branchial cleft cysts are the most common.[3]

Thyroglossal cysts and fistulas are similar defects located in or near the midline of the neck; they may extend to the base of the tongue. A characteristic feature is vertical motion of the mass with swallowing and tongue protrusion. Cysts in the tongue base may also be differentiated from an undescended lingual thyroid by radionuclide scanning.

Branchial cysts, sinuses, and fistulas may develop from parts of the second pharyngeal groove, the cervical sinus, or the second pharyngeal pouch that fail to obliterate.

Branchial cleft anomalies are remnants of the four paired embryonic branchial arches, clefts, and pouches. These lesions may present as sinuses, fistulas, or cartilaginous rests in infants but more commonly present as cysts in older children, adolescents, and adults.

The important fact is that the first branchial cleft sinus presents anterior to the ear and connects with the eustachian tube. The most frequently observed anomaly involves the second branchial cleft remnant, which may present clinically anywhere along the anterior border of the sternocleidomastoid muscle from the angle of the jaw to the lower third of the neck (as a sinus tract).

External branchial sinuses are uncommon and almost all result from failure of the second pharyngeal groove and the cervical sinus to obliterate. The sinus typically opens along the anterior border of the sternocleidomastoid muscle in the inferior third of the neck. Anomalies of the other pharyngeal grooves occur in approximately 5-7% of cases. External branchial sinuses are commonly detected during infancy because of the discharge of mucous material from them.

BRANCHIAL FISTULA

An abnormal canal that opens internally into the tonsillar sinus and externally in the side of the neck is a branchial fistula. This canal results from persistence of parts of the second pharyngeal groove and second pharyngeal pouch. The fistula ascends from its opening in the neck through the subcutaneous tissue and platysma muscle to reach the carotid sheath.

Embryologically the third and fourth branchial clefts develop into the lower and upper parathyroid glands, respectively. Any aberration in their development may be part of a pharyngeal pouch deficiency syndrome characterized by absent parathyroids and an absent or depleted thymus DiGeorge's syndrome, producing an immunosuppressive syndrome (T-cell depletion), hypocalcemia, and seizures. Aortic arch vascular anomalies may also coexist. DiGeorge syndrome occurs because the third and fourth pharyngeal pouches fail to differentiate into the thymus and parathyroid glands. This is the result of a breakdown in signaling between pharyngeal endoderm and adjacent neural crest cells. In most cases of DiGeorge syndrome, there is a microdeletion in the q11.2 region of chromosome 22, HIRA and UFDIL gene mutation, and neural crest cell defects.

Infants with DiGeorge syndrome are born without a thymus and parathyroid glands and have defects in their cardiac outflow tracts. Clinically, the disease is characterized by congenital hypoparathyroidism, increased susceptibility to infections (from immune deficiency, specifically defective T-cell function), anomalies of the mouth, low-set ears, nasal clefts, thyroid hypoplasia, and cardiac abnormalities (defects of the arch of the aorta and heart).[4]

Paired branchial clefts and arches develop early in the fourth gestational week. The first cleft and the first, second, third, and fourth pouches give rise to adult organs. The embryologic communication between the pharynx and the external surface may persist as a fistula. A fistula is seen most commonly with the second branchial cleft, which normally disappears, and extends from the anterior border of the sternocleidomastoid muscle superiorly, passes inward through the bifurcation of the carotid artery, and enters the posterolateral pharynx just below the tonsillar fossa. In contrast, a third branchial cleft fistula passes posterior to the carotid bifurcation. The branchial cleft remnants may contain small pieces of cartilage and cysts, but internal fistulas are rare. A second branchial cleft sinus is suspected when clear fluid is noted draining from the external opening of the tract at the anterior border of the lower third of the sternomastoid muscle. Rarely, branchial cleft anomalies occur in association with biliary atresia and congenital cardiac anomalies, an association that is referred to as Goldenhar's complex.

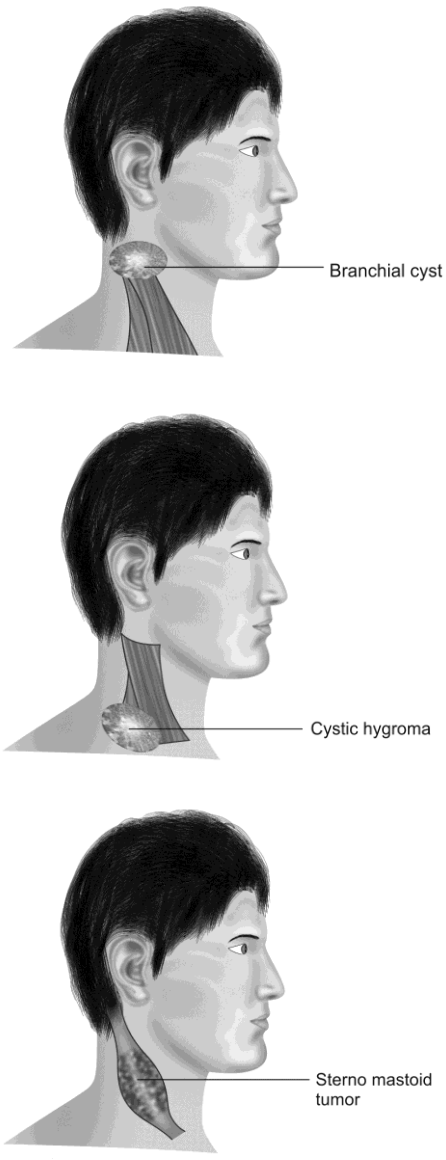
Thyroglossal duct cysts represent the vestigial remainder of the tract of the descending thyroid gland from the foramen cecum, at the tongue base, into the lower anterior neck during fetal development.[5] They present as a midline or paramedian cystic mass adjacent to the hyoid bone. After an upper respiratory infection, the cyst may enlarge or become infected. Surgical management of a thyroglossal duct cyst requires removal of the cyst, the tract, and the central portion of the hyoid bone (Sistrunk procedure), as well as a portion of the tongue base up to the foramen cecum.

Thyroglossal duct anomalies are midline lesions that originate at the base of the tongue at the foramen caecum and pass through the central portion of the hyoid bone. The thyroglossal duct usually obliterates in the sixth intrauterine week. Persistence of this fetal structure postnatally results in a midline swelling over the hyoid bone that moves with deglutition. Infection is the most common complication. A sinus tract may develop as a result of infection. The differential diagnosis of upper midline neck masses includes inflamed submental lymph node, pretracheal dermoid cyst, or ectopic thyroid tissue. The lymph node usually responds to antibiotics, is movable, and is separate from the hyoid bone. Ectopic thyroid is rare, does not get infected, and always appears solid on ultrasonography.

Cystic hygroma occurring as a result of sequestration or obstruction of developing lymph vessels. Although the lesion can occur anywhere, the most common sites are in the posterior triangle of the neck, axilla, groin, and mediastinum. The cysts are lined by endothelium and filled with lymph.

Congenital anomalies of the lymphatic system are not that uncommon. Hygromas are believed to arise from parts of a jugular lymph sac that are pinched off or from lymphatic spaces that fail to establish connections with the main lymphatic channels. There may be diffuse swelling of a part of the body-congenital lymphedema. This condition may result from dilation of primordial

lymphatic channels or from congenital hypoplasia of lymphatic vessels. More rarely, diffuse cystic dilation of lymphatic channels involves widespread portions of the body. In cystic hygroma, large swellings usually appear in the inferolateral part of the neck and consist of large single or multilocular, fluid-filled cavities. Hygromas may be present at birth, but they often enlarge and become evident during infancy. Most hygromas appear to be derived from abnormal transformation of the jugular lymph sacs. Hygromas are believed to arise from parts of a jugular lymph sac that are pinched off or from lymphatic spaces that fail to establish connections with the main lymphatic channels.



Embryologically cystic hygroma presents as developmental lymphangioma is derived from the primitive embryonic jugular venolymphatic sacs. It usually may present as a mass at birth with rarely the mass extending into the axilla or mediastinum. The cystic form is frequently intimately adherent to vital structures (e.g., vagus nerve, phrenic nerve) that should not be sacrificed, since this lesion is benign.

TORTICOLLIS OR WRY NECK

Torticollis or wry neck may present as a hard, spindle-shaped, fibrous tumor or swelling within the sternocleidomastoid muscle. It may be detected in the first few weeks of life usually as a result of birth injury. It occurs commonly following breech delivery and may be associated with eventration of the diaphragm and Erb's palsy on the same side. Muscular torticollis is the most common type and usually occurs at birth from a suspected injury to the sternocleidomastoid muscle. Large infants who have had difficult vertex deliveries are at special risk. Stretching of the neck during delivery results in tearing and bleeding within the sternocleidomastoid muscle. The blood is contained within its own fascial compartment, resulting in increased pressure. The increased pressure further damages the muscle, resulting in an area of ischemia that secondarily becomes replaced by fibrous tissue, contracts, and produces torticollis.

The clinical presentation includes a hard mass within the sternocleidomastoid muscle, ipsilateral facial hemihypoplasia, plagiocephaly, head turned away from the side of the mass, and, occasionally, ipsilateral trapezius atrophy.

HEMANGIOMAS

Developmental vascular anomalies may occur as isolated defects or as part of a syndrome. They can be separated into two major categories: hemangiomas and malformations. Hemangiomas are proliferative lesions of vascular endothelium that predictably enlarge and then spontaneously involute. Hemangiomas are the most common tumor of infancy.

Cavernous hemangiomas: They are deeply situated lesions and appear as acystic, firm, or compressible masses in the neck and the overlying skin may appear normal in color or have a bluish hue. Cavernous hemangiomas progress from a growth phase to a stationary phase to a period of involution. These lesions are as likely to regress as capillary hemangiomas, and the outcome cannot be predicted from size or site of involvement.[6] Conservative approach is usually sufficient. If involvement of underlying structures is suspected, appropriate radiologic studies should be performed for elucidation. Rarely, these lesions impinge on vital structures, interfere with functions such as vision or feeding, cause grotesque disfigurement because of rapid growth, or are associated with life-threatening complications such as thrombocytopenia and hemorrhage. Kassabach Merit Syndrome can be a clinical

possibility. Hemangiomas associated with a generalized bleeding disorder caused by the trapping of platelets within them which produces a profound thrombocytopenia.

DERMOID CYSTS

These usually occur at sites of embryological fusion. These may be in the midline. A dermoid cyst in the neck may be mistaken for a thyroglossal cyst although it will not move on swallowing or protrusion of the tongue. A common site is the external angular dermoid cyst in the eyebrow area at the outer angle of the eye. Occasionally there may be a dumbbell extension intracranially. They occur if ectodermal cells become buried beneath the skin surface during development. An inclusion dermoid cyst may similarly arise secondary to trauma.

Neck masses in children can also be caused by a benign or malignant tumor. Differential diagnosis for any pediatrician is important because these conditions can be easily confused with more malignant lesions. Primary tumors that arise in the neck in the pediatric age group include thyroid and parathyroid tumors, Hodgkin's disease, non-Hodgkin's lymphomas, cervical neuroblastoma, rhabdomyosarcoma, leukemia, teratomas, neurofibromas, submandibular tumors, parotid tumors, Histiocytosis-X to name a few.

REFERENCES

1. Filston HC. Head and neck: Sinuses and masses. In Holder, T. M., and Ashcraft, K. W. (Eds.): Pediatric Surgery. Philadelphia, W. B. Saunders, 1981, p. 1062.
2. Telander RL, Deane SA. Thyroglossal duct and branchial cleft cysts and sinuses. Surg Clin North Am. 1977;57:779.
3. Hinrichsen K. The early development of morphology and patterns of the face in the human embryo. Adv Anat Embryol Cell Biol. 1985;98:1-79.
4. Goldmuntz E. DiGeorge syndrome: New insights. Clin Perinatol. 2005;32:963.
5. Clark OH, Okerlund MD, Cavalieri RR, Greenspan FS. Diagnosis and treatment of thyroid, parathyroid, and thyroglossal duct cysts. J Clin Endocrinol Metab. 1979;48:983.
6. Yousem AL, Wick MR, Singh G, Katyal SL, Manivel CJ, Mills SE, Legier J. So-called sclerosing hemangioma of lung. Am J Surg Pathol. 1988;12:582-90.

DOI: 10.5455/2320-6012.ijrms20131103
Cite this article as: Rasool Z, Hassan AU. Congenital neck masses: embryological and anatomical perspectives. Int J Res Med Sci 2013;1:329-32.