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A Review on a Rare Congenital Disorder: Congenital Midline Cervical Cleft

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ABSTRACT

Congenital Midline Cervical Cleft (CMCC) is a very rare congenital disorder of the anterior neck.
Even though the embryologic mechanism that accounts for the congenital midline cervical cleft is not firmly established, most researchers believe that it involves a failure of the branchial arches to unite in the midline. The main characteristics of the defect are: (1) a caudally located fistula opening; (2) intermittent serous fluid discharge in the early neonatal period; (3) nipple-like protuberance of the cleft in the superior aspect; (4) a widened scar and minimal neck contracture in later life. Diagnosis is based on these features found at birth. The method of choice is surgical intervention in the form of Z-plasty technique. Early treatment is significant to prevent cervical contractures, scarring, and to achieve beneficial cosmetic results.

Keywords: Congenital midline cervical cleft, Midline cervical cleft, Congenital neck anomalies, Surgical excision, Z-plasty

DEFINITION

Congenital Midline Cervical Cleft (CMCC) is a very rare congenital anomaly of the anterior neck. It appears to occur as a result of a failure of fusion of the paired second branchial arches in the midline during embryogenesis [1].

INCIDENCE

In 1848, Luschka described the first case of CMCC, while Bailey reported the first description of this abnormality in 1924. By 2014 only 205 cases had been documented [2-4].

A female to male ratio of 2:1 is reported, with a sporadic presentation [5]. Especially Caucasian females appear to be the most affected [6]. The age of presentation ranges from birth to 23 years [7].

PATHOGENESIS

The embryo pathogenesis of CMCC has not been established, although several theories have been proposed, the most widely accepted being the impaired fusion of the first or second branchial arches during the third and fourth weeks of embryonic development [8].

The CMCC consists of epidermis, a mucosal surface skeletal muscle that is superficial to glandular tissue and platysma. In 1949, Ombredanne et al. suggested that pressure from the pericardial roof on the developing branchial arches results in pressure necrosis and scarring [9]. Oostrom et al. [10] hypothesized that rupture of a pathological adhesion between the epithelium of the cardiohepatic fold with that of the ventral part of the first branchial arch leads to tissue ischemia, again with localized necrosis and scarring.

However, these theories do not explain the consistent anatomy of the CMCC or the presence of skeletal muscle and glandular tissue [6].

Furthermore the atypical development of the first branchial arch may also provide a possible explanation for the histological findings. The branchial arches start to form on day 22 in the human embryo. A horizontal cleft divides the first arch into maxillary and mandibular processes each side at the midline. On day 26 the mandibular process fuses. A delay in this process could cause a deposition of ectodermal cells with underlying mesodermal cells in the ventral side of the neck. These would continue to modify and form skeletal muscle such as tongue derivative, salivary glandular tissue and a mucosal surface, thus resulting in the CMCC defect [3,6].

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Due to this second theory hair follicles or sweat or sebaceous glands are absent [1]. It may be concluded that the skin protuberance could be a vertical excrescence of tongue muscle, since the fibrous cord is connected to the fibrous median septum of the tongue base; salivary glands in the sinus tract and mucoid discharge contribute to the theory [11].

Surgical intervention should be performed at an early stage, since the traction of the cord on the mandibular bone during development may produce abnormalities such as exostosis, micrognathia and torticollis [8]. Congenital midline cervical cleft may also be associated with other malformations in the region, such as thyroglossal duct cyst, ectopic bronchogenic cyst, cleft tongue, lower lip and mandible or cleft sternum, hypoplasia or absence of the hyoid bone, and can also be associated with cardiac anomalies [1].

DIAGNOSIS

Diagnosis is based on the clinical features of CMCC **(Table 1)** and the main characteristic is a protruding lesion at the midline of the anterior region of the neck, between the chin and the suprasternal notch **(Figure 1)**. The lesion is superficial and the superficial muscles of the neck remain intact [3].

Table 1. Main features of congenital midline cervical cleft [13].

The cardinal features of congenital midline cervical cleft are:
A caudally located fistula opening
Intermittent serous fluid discharge in the early neonatal period
Nipple-like appearance of the cleft in the superior aspect
A widened scar and minimal neck contracture in later life

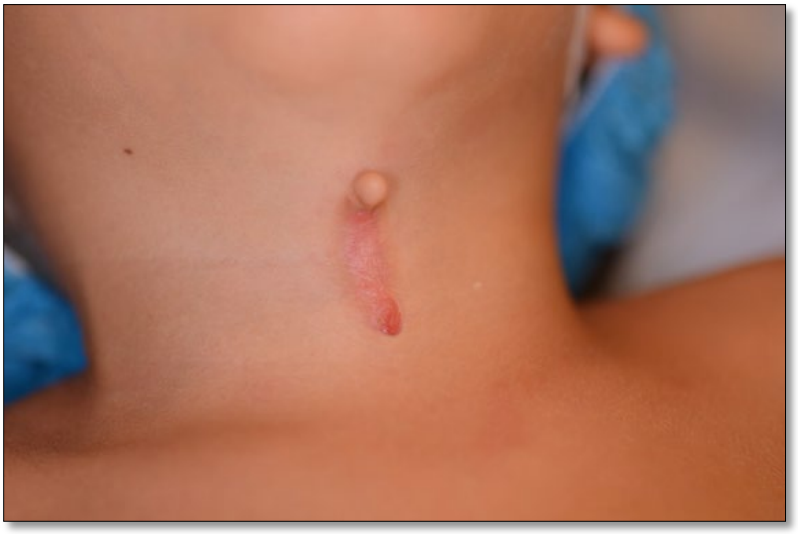

Figure 1. 3 year old boy with CMCC located in the ventral midline of his neck. The superior part shows a skin tag leading to a short mucosa-like raw surface.

It is not a true cleft, because there exists no skin gap. A mucosal surface bisects a skin tag and a short sinus (normally about 1 cm in length). CMCC constitutes a variation of the cleft category number 30 of the Tessier classification system of craniofacial defects [2,12,13].

The lesion extends caudally as a longitudinal cleft lined by a reddened and desquamative epithelium. Generally, a mucus-secreting opening of the fistulous tract exists in the most caudal portion of the cleft **(Figure 1)**. The seromucinous discharge resolves gradually the first months of infancy [2]. Over time the cleft heals and a longitudinal scar is formed, developing the formation of web, which causes contracture of the neck, limits neck mobility, particularly extension, or torticollis [7,14].

Additionally, palpation reveals a subcutaneous fibrous cord that extends to the entire cleft or part of it, from the submental region to the suprasternal notch [3]. It is also important to probe the sinus and visualize the blind end by means of radiography.

Histologic examination has revealed stratified keratinized squamous epithelium lacking skin appendages such as sweat or sebaceous glands. The dermis has alternating hypertrophy and atrophy and there is a mild lymphocytic inflammatory infiltrate in the subcutaneous tissues [15].

An associated sinus tract is lined by a pseudostratified epithelium with seromucinous salivary glands; however, there have been several reports of respiratory epithelium and bronchial glands associated with the sinus tract **(Figure 2)** [16].

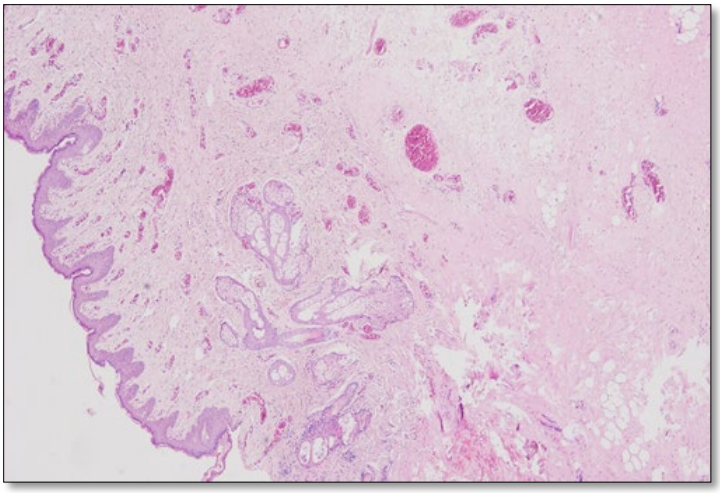

Figure 2. Histologic presentation of the cleft. Stratified squamous epithelium covers the dermis with adnexes.

RADIOLOGICAL ASPECT

Ultrasonographic examination is significant to exclude associated abnormalities of the thyroid gland and other related anomalies. The regional site of the sinus tract is important in distinguishing the CMCC from a thyroglossal fistula, since the tract extends caudally in CMCC and cranially in the thyroglossal fistula [11].

Previous to the widespread use of ultrasound and MRI, radiographs of the neck typically demonstrated bony spurring at the mandible in advanced cases due to traction from the underlying fibrous cord. Ultrasound of the neck shows a non-vascular, blind-ending sinus tract from the skin surface. Mostly the thyroid gland is normal.

MRI defines the lesion, its course and excludes other differential considerations such as thyroglossal duct cysts or branchial cleft anomalies **(Figure 3)**. MRI usually shows skin thinning and a peripherally - enhancing, T1 hypointense and T2 hyperintense linear tract without disruption of any bony or cartilaginous structures or involvement of the thyroid gland **(Figures 3-5)** [17].

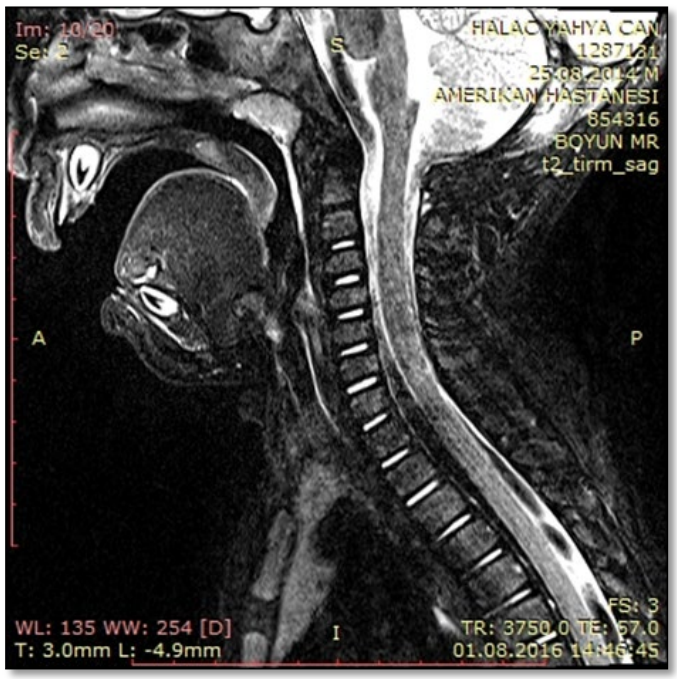

Figure 3. Sagittal T2-weighted magnetic resonance imaging of the patient's neck demonstrating the cleft on the ventral side.

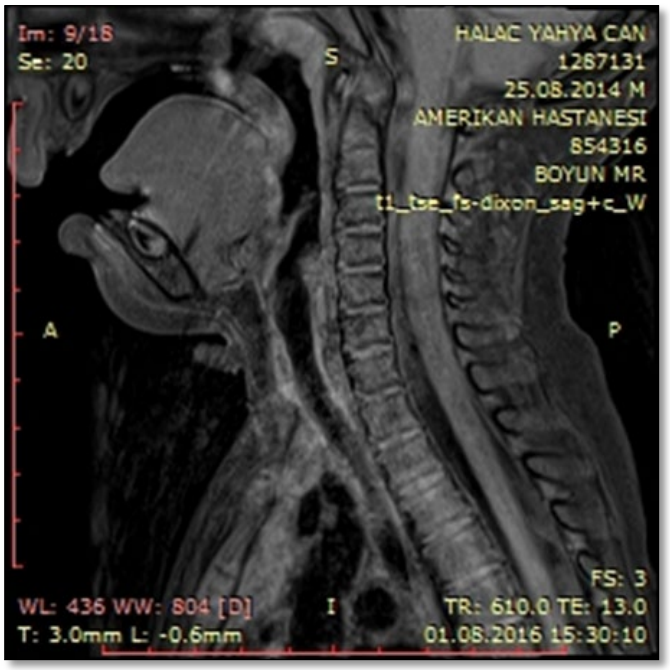

Figure 4. Sagittal T1-weighted MRI of the defect. The nipple-like protuberance is located between the chin and the suprasternal notch.

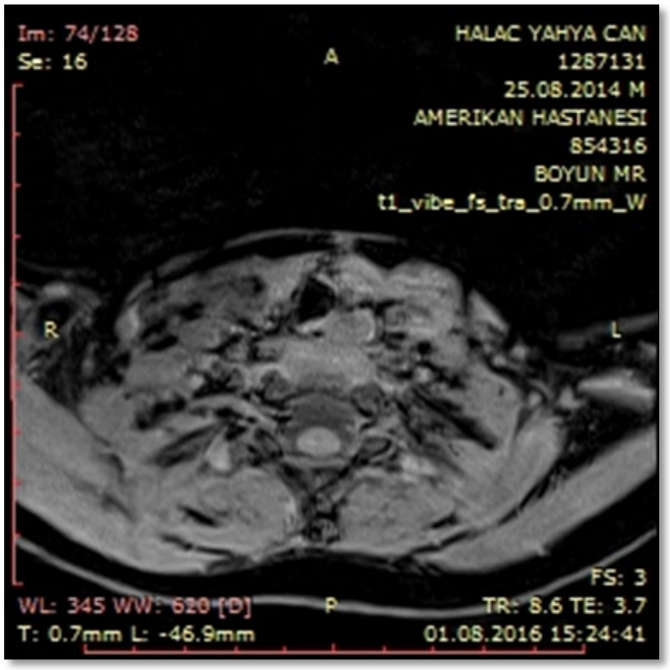

Figure 5. Patient's neck is shown in axial plane with MRI. Note the cleft on the anterior side of the neck.

DIFFERENTIAL DIAGNOSIS

Differential diagnosis includes thyroglossal duct cysts or branchial cleft anomalies **(Table 2)**. A thyroglossal duct cyst characteristically appears cystic, although the midline T2 hyperintense lesion have faint peripheral enhancement **(Figure 3)**. A thyroglossal duct cyst is classically much higher in the neck, being more midline when related to the hyoid and more lateral when located in the infrahyoid neck. Moreover, a thyroglossal duct cyst does not usually have an associated sinus tract unless it is superinfected. 2nd or 3rd branchial cleft cysts or pyriform sinus fistulae are typically off midline in the lateral neck [17].

Table 2. Differential diagnosis in terms of radiological characteristics for congenital midline cervical cleft [17].

	Congenital midline cervical cleft	**Thyroglossal Duct Cyst**	**Second Branchial Cleft Anomaly**
X-ray	Sometimes bony mandibular spur is seen	Generally no findings	Generally no findings
Ultrasound	Blind ending sinus tract in the midline	Anechoic or hypoechoic cyst near the hyoid bone located in the midline	Anechoic or hypoechoic cyst located posterolateral to submandibular gland and anterior to sternocleidomastoid muscle
Magnetic Resonance Imaging (MRI)	T1 hypointense, T2 hyperintense tract with peripheral expansion	Cyst is hypointense on T1 and hyperintense on T2, cyst wall enlarges if infected	Cyst is hypointense on T1 and hyperintense on T2, cyst wall enlarges if infected
Computed Tomography (CT)	Not commonly visualized, blind ending sinus tract can be seen	Low-attenuating cyst, mild peripheral expansion	Low-attenuating cyst, wall is not expanded, may expand in case of infection

TREATMENT

CMCC is treated with surgical resection and proper reconstruction to avoid scarring, to prevent cervical contractures and to cure other associated congenital abnormalities.

A timely treatment of CMCC is essential to prevent changes in the growth of the lower third of the face, mandibular development and extension of the neck [18]. Reconstruction of the defect needs surgical maneuvers other than primary closure. Primary closure would lead to an unsightly scar and a vertical scar band that would cause restriction of neck movements. Consequently, Z-plasty is one of the surgical procedures used to close the defect. Single, double or multiple Z-plasty is the treatment of choice for CMCC **(Figure 6)**.

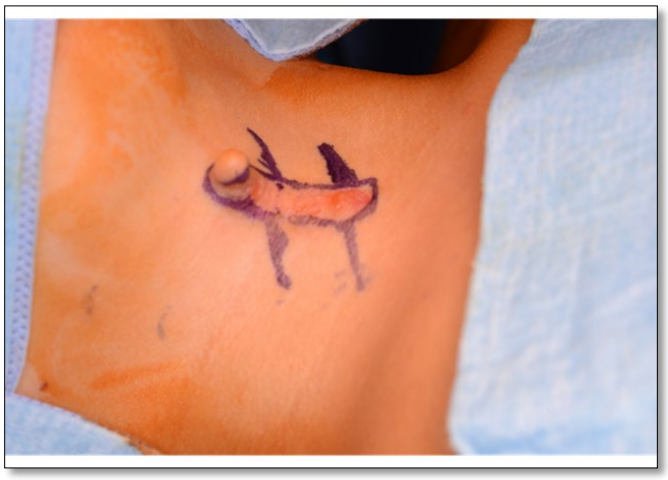

Figure 6. Surgical markings for resection of the lesion and Z-plasty.

According to long-term functional results of congenital midline cervical cleft, Z-plasty closure have been reported to be satisfactory with improvement in vertical neck movement **(Figure 7)**. Cosmetic results are more variable. Horizontal scars often remain thin but widening of the oblique limbs of the Z-plasty scar has been described [18].

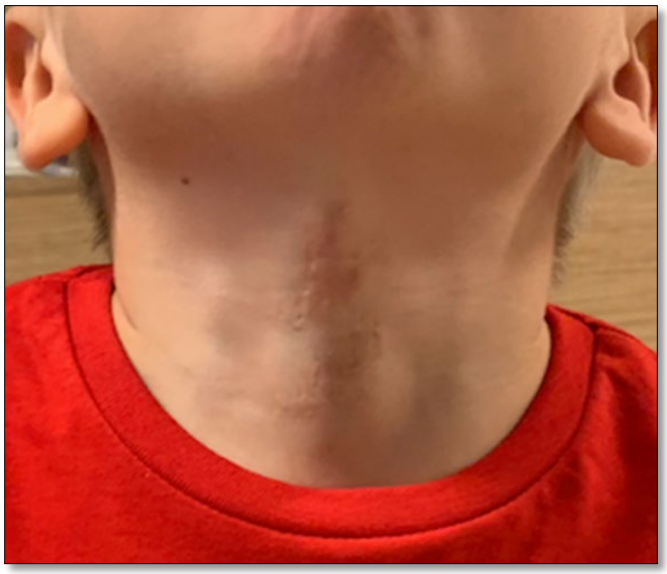

Figure 7. Long-term post-operative results of Z-plasty. Note that the patient is able to hyperextend his neck without any restriction.

Nevertheless more advanced cases having hypoplasia of mandible, absent hyoid and/or thyroid cartilage or other supporting structures of the neck warrant extensive teamwork between plastic surgeon, head and neck surgeon, maxillofacial surgeon, which might need to be supported by a psychologists, speech therapist, paediatrician and very dedicated nursing care [2].

Therefore a correct earlier recognition of the lesion and appropriate surgical management are indispensable to avoid long-term complications [2].

CONCLUSION

CMCC is an uncommon congenital malformation which diagnosis is made on clinical examination. The main characteristic is a midline cutaneous protuberance over a linear vertical erythematous area. A fibrous band located under this lesion usually extends to the manubrium and a fistulous opening can be noticed at the caudal end. Surgical excision and reconstruction of all pathologic tissues with multiple Z-plasty reconstruction must be performed immediately in order to limit the risk of recurrence and avoid limitation of extension of the neck **(Figure 7)**.

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