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CASE REPORT

Congenital midline cervical cleft: Clinical approach to a congenital anterior neck defect

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ABSTRACT **Numerous malformations can affect the anterior part of the neck presenting at birth as a real diagnostic challenge for the pediatrician or the primary care physician who initially evaluate the baby. Congenital midline cervical cleft represents a rare defect of the midline neck, which is sometimes wrongly diagnosed as a thyroglossal duct anomaly, dermoid cyst, branchial cleft anomaly or "birthmark". A prompt clinical diagnosis and surgical treatment during early infancy are essential to ensure both functional and aesthetic outcome. We report a case of a female neonate with a midline cervical cleft diagnosed immediately after birth. The main features of other congenital anomalies of the anterior neck are also discussed referring to their embryologic origin.**

Key Words: craniofacial malformation, mentosternal dysraphia, midline cervical cleft, pediatrician

INTRODUCTION

Congenital midline cervical cleft (CMCC) is a rare congenital anomaly of the anterior part of the neck (Origlio et al. 1966; McInnes et al. 2012), also known as mentosternal dysraphia (Origlio et al. 1966). The incidence of CMCC among the other congenital cervical malformations ranges between 1.7–2.0% (Kokodkar et al. 2013). One of the largest reported series includes 10 patients aged 4 to 27 years who were observed at the "Istituto di Chirurgia Plastica e Ricostruttiva dell'Università degli Studi di Milano" (Origlio et al. 1966). Several hypotheses have been postulated concerning its occurrence from an embryological point of view: exteriorization of a thyroglossal duct remnant or increased pressure in the cervical area from the pericardial roof in the developing embryo, but the most widely accepted theory supports an impaired midline fusion of the branchial arches (Eastlack et al. 2000; Mlynarek et al. 2003; McInnes et al. 2012). Throughout normal development, the branchial arches grow medially and merge in a cephalo-caudal direction with the first arch, closing before the second, and the lower arches follow sequentially. Prior to fusing, mesodermal tissue migrates between the arches, pushing ectoderm outward to flatten the ventral furrow (Maddalozzo et al. 1993). Mentosternal dysraphies represent the passage between the craniofacial sector and the other districts of the body (Van der Meulen et al. 1990).

The typical presentation of CMCC shows:

- a superior nipple-like skin tag
- a reddened weeping strip of atrophic skin occurring at any level between the chin and the sternal notch
- a sinus tract at the caudal end which can discharge mucoid material (Gardner and Moss 2005; Kokodkar et al. 2013)

A subcutaneous fibrous cord is always present and may represent the only feature in partial clinical manifestations. The lack of protrusion of the chin, appearing flat and wide, sometimes with midline notch, may be observed in CMCC. In these cases, radiological examinations can show an underdeveloped mandible (Origlio et al. 1966). Besides this classical presentation, a few more complex cases have been reported. A case with a cleft of the lower lip and jaw, which extended up to the cricoid cartilage has been described (Stewart 1935; Origlio et al. 1966). Bifidity of tongue was associated. A case with a large cleft of the lower lip and mandibular arch has been reported (Davis 1950; Origlio et al. 1966). The hyoid bone, the cricoid cartilage and the manubrium of sternum were absent. A case described (Morton and Jordan 1935; Origlio et al. 1966) had a large cleft of the inferior lip and the mandible along with a sternal and tongue cleft. Therefore, in the presence of CMCC, other midline defects need to be excluded: cleft of the lower lip, mandible, chin, tongue and sternum as well as midline abdominal web or scar-like raphe, midline hemangioma and congenital heart lesion (Hirokawa et al. 2003; Kokodkar et al. 2013). Moreover, thyroglossal duct cysts, bronchogenic cysts and dermoid cysts may be associated with CMCC (Mlynarek et al. 2003; Vure et al. 2009) and should be excluded as well.

With time the cleft heals and a scar is formed causing contracture of the neck and limiting the neck mobility, mainly the extension. For this reason surgical treatment is advised (Sinopidis et al. 2012). Most authors recommend a Z-plasty to correct anterior cervical contractures (Cochran et al. 2006; Kokodkar et al. 2013). The Z-plasty consists of transposing two triangular skin flaps, interchanging one for another, with the purpose of obtaining a gain in length of a linear scar contracture. Long-term functional results of the congenital midline cervical cleft Z-plasty have been reported as satisfactory with an improvement in vertical neck extension.

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However, no general consensus exists about the appropriate age for elective surgical correction of midline cervical cleft (Kokodkar et al. 2013).

We describe a case of a baby with midline cervical cleft promptly recognized at birth, reporting the indication and the timing of correction. The purpose of the present paper is to inform the physician about the differential diagnosis to be considered when evaluating a baby with a congenital anterior neck defect. A summary table is also provided.

CLINICAL REPORT

The proband is the second child born to a 32-year-old woman. Both parents were healthy and non-consanguineous and no drugs had been taken during the gestational period. The pregnancy was uneventful. The baby, a female, was delivered by caesarean section at 39 weeks of gestation with an Apgar score of 9 at the 1st and 10 at 5th minute. Birthweight was 3465 g (50–75th centile), length 49 cm (25–50th centile) and head circumference 37.2 cm (>97th centile). At birth a 2-cm long and 0.5-cm wide strip of atrophic skin extending from the level below the hyoid bone to the suprasternal notch was observed. On palpation, a midline notch of the lower border of the mandible was evident. There was a nipple like skin swelling of 3×2 mm at the cranial end and a sinus at the caudal end (Fig. 1). A moderately retractile subcutaneous cord, ranging from the mandible to the manubrium, was noted when the neck was hyperextended, along with a superficial cleft of the inferior lip. She was otherwise healthy: in particular, no respiratory distress, dysphagia, reflux or hearing impairment were observed. An ultrasound scan of the neck was performed to exclude the association with other neck abnormalities. The echocardiography and the abdominal ultrasound examination were normal and no other midline defect was found. The otolaryngological evaluation confirmed the diagnosis of isolated midline cervical cleft. A clinical follow-up was planned. At 6 months of age, the child is doing well, clinically unchanged, with no apparent neck extension impairment. The surgical treatment with a Z-plasty will be planned prior to school age in order to allow a better extension of the neck and ensure a functional and aesthetic outcome.

DISCUSSION

Congenital midline cervical cleft represents a distinctive anomaly and its diagnosis can be made on physical examination (Kokodkar et al. 2013) although it can be overlooked in the differential diagnosis of similarly presenting, more common neck anomalies (Tsukono et al. 2002; McInnes et al. 2012).

Firstly, thyroglossal duct anomalies, the most common midline cervical defects, need to be excluded (Foley and Fallat 2006; Waldhausen 2006). In early gestation the thyroid gland originates in the base of the tongue and later it is caudally displaced in the neck. During this process the median anlage elongates proximal to the descending gland forming the thyroglossal duct. It normally obliterates by the fifth week of gestation before the formation of the mesodermal anlage of the hyoid bone. Failure of the thyroglossal duct to obliterate can result in its persistence at birth (Foley and Fallat 2006). The thyroglossal duct remnants usually present as cystic masses and up to 25% of lesions similar to a draining sinus tract. Because of its relationship to the hyoid bone the cyst moves cranially with swallowing and the protrusion of the tongue, although it is difficult to observe in babies (Foley and Fallat 2006). The cranial midline location and the close association with the hyoid bone are the typical features of this anomaly (Foley and Fallat 2006). Differently, CMCC has a midline caudal location (Mendis and Moss 2007) and no relation to the hyoid bone. Moreover, the sinus of CMCC is generally caudally oriented, whereas a sinus associated with a thyroglossal duct cyst tends to be cranially directed (McInnes et al. 2012). It is important to differentiate these two entities as treatment of a thyroglossal duct cyst involves the dissection of the duct and the simultaneous removal of the central part of the hyoid bone. However, the association between thyroglossal cysts and CMCC may occur (McInnes et al. 2012). For this reason an ultrasound scan of the neck is mandatory.

Branchial cleft anomalies, the most common congenital head and neck lesions, have to be considered in differential diagnosis (Waldhausen 2006; Geddes et al. 2013). The branchial structures develop by the end of the fourth week and consist of four well-defined pairs of branchial arches prominent in the lateral profile and two additional rudimentary arches. Arches are separated from each other externally by ectoderm-lined branchial clefts and internally

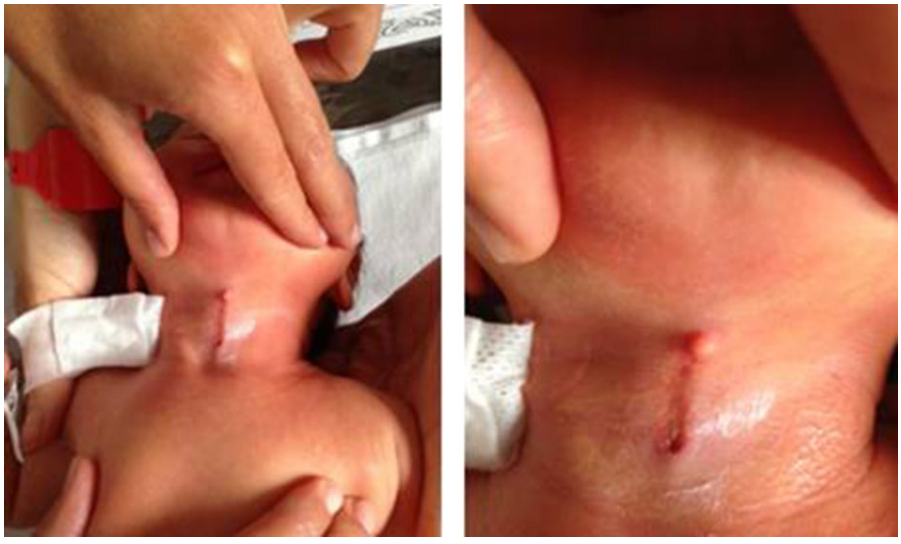

Fig. 1 Clinical features of congenital midline cervical cleft (CMCC) in our patient. Note the superior nipple-like skin tag, the reddened weeping strip of atrophic skin from the level below the hyoid bone to the suprasternal notch and the sinus tract at the caudal end.

Table 1 Clinical features of congenital anterior neck defects

CMCC	– Midline caudal location – It consists of three anatomic parts: a superior nipple-like skin tag, a strip of atrophic skin between the chin and the sternal notch a sinus tract at the caudal end – A subcutaneous fibrous cord is always present and may represent the only feature in partial clinical presentations – Lack of protrusion of the chin which appears flat and wide, sometimes with hints of cleft. – Commonly no relation to the hyoid bone Exclude: – other midline defects including cleft of lower lip, mandible, chin, tongue and sternum, midline abdominal web or scar-like raphe, midline hemangioma and congenital heart lesion – thyroglossal duct cysts and bronchogenic cysts could present simultaneously with CMCC
Thyroglossal duct anomalies	– Midline cranial location – Commonly presenting as cystic masses with a draining sinus tract in up to one-quarter of lesions – Relationship to the hyoid bone – Moving cranially with swallowing and the protrusion of the tongue – Cranially directed sinus – Could present simultaneously with CMCC
Branchial anomalies The second branchial apparatus ones represent up to 95%.	– Located in the lower, anterior-lateral region of the neck – Bilateral involvement uncommon – Fistulae have a chronic drainage from an opening along the anterior border of the sternocleidomastoid muscle. Exclude: – BOR syndrome
Dermoid cysts	– Midline location when they occur in the anterior neck – Painless, superficial, subcutaneous palpable masses – Do not move with swallowing

BOR, Brachiootorenal; CMCC, Congenital midline cervical cleft.

by endoderm-lined pharyngeal pouches. Branchial anomalies arise from the incomplete obliteration of cleft and pouches and they might present as cyst, sinus or fistulae. Among the branchial anomalies the second branchial apparatus lesions are the most common and account for up to 95% of these abnormalities. Differently from CMCC, they are usually located in the lower, anterior-lateral region of the neck. A bilateral involvement is uncommon. Fistulae have a chronic drainage from an opening along the anterior border of the stern mastoid muscle. Anomalies of the third and fourth arch, rarely described, appear similar externally: only their internal opening allows the definition of the origin. Ultrasonography, computed tomography and magnetic resonance imaging can help to characterize these anomalies and to evaluate the surrounding structures. The first cleft remnant, presents as a cyst, sinus or fistula somewhere between the external auditory canal and the submandibular area (Waldhausen 2006). Branchial fistulae may be part of a complex association, the brachiootorenal (BOR) syndrome, which includes auricular malformation, preauricular pits, inner ear anomalies, deafness and renal abnormalities (Chen et al. 1995).

Finally, CMCC are easily differentiated from dermoid cystsm, which represent up to 25% of midline cervical anomalies (Geddes et al. 2013; LaRiviere & Waldhausen 2012; Pryor et al. 2005). A dermoid cyst is a benign neoplasm derived from both endoderm and mesoderm. A keratinizing squamous epithelium is typically present together with dermal derivatives such as hair follicles, smooth muscle, sweat and sebaceous glands, and fibroadipose tissue (Pryor et al. 2005). These cysts present as painless, superficial, subcutaneous palpable masses. When they occur in the anterior neck they are usually located in the midline and close to the hyoid bone. For this reason, they may be confused with thyroglossal duct cysts. Because of their superficial location and lack of mesodermal attachments, dermoid cysts do not move with swallowing (Geddes et al. 2013).

The most important features of the reported anomalies are shown in Table 1.

CONCLUSION

Congenital midline cervical cleft is one of the rarest congenital anomalies of the neck and although it presents with well-defined features, it is often unrecognized at birth. CMCC has to be considered, when assessing a child with a midline cervical lesion, and differentiated from other more common neck anomalies. An early

recognition at birth and a structured diagnostic assessment are necessary to ensure a favorable outcome. CMCC requires a clinical and surgical follow-up which has to be planned after birth. The surgical correction of this malformation is advisable in order to avoid secondary deformities of the jaw and sternum and neck extension impairment. Although the exact timing of surgery is still a matter of debate (Kokodkar et al. 2013) early surgical excision with Z-plasty has to be taken into consideration (Farhadj et al. 2012).

DISCLOSURE

None.

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