Congenital Midline Cervical Cleft: A Variant of Tessier Number 30 Cleft Causing Micrognathia

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Abstract

Congenital midline cervical cleft is a rare and generally isolated malformation of the ventral neck region with no clear etiology established. Mandibular deformities, such as micrognathia, could be considered as a consequence of a cleft cervical contracture. Complete surgical excision of the subcutaneous fibrous cord at an early age is the primary treatment modality, minimizing growth development problems on surrounding affected tissue. The aim of this study is to describe the clinical, surgical, and histological findings in a female child with congenital midline cervical cleft along with a relevant literature review. Three years follow-up after surgery exhibited satisfactory functional and cosmetic results.

Introduction

Congenital midline cervical cleft (CMCC) is a rare developmental anomaly of the ventral neck (Ayache et al., 1997; Bajaj et al., 2004; McInnes et al., 2012) with an incidence ranging from 1.7% to 2% among congenital cervical malformations, including thyroglossal duct anomalies, dermoid cysts, and ranula (Crippa et al., 2015; Achard et al., 2016; Bahakim et al., 2018).

It represents a variant of Tessier 30 cleft (Erçöçen et al., 2002; Jakobsen et al., 2012; Sinopidis et al., 2012; Saha et al., 2013; Eom et al., 2014; Puscas, 2015; Bahakim et al., 2018). It may be of variable length and width and occur in the midline anywhere between the symphysis of the mandible and the suprasternal notch (Cochran et al., 2006); however, it is not considered a true cleft because it does not involve a gap between adjacent skin flaps (Stephen et al., 1989).

It was first described by Luschka in 1848 (Derbez et al., 2004; Goldfisher et al., 2015; Puscas, 2015). Bailey made the first English description in 1925 (Achard et al., 2016). Since then, nearly 120 articles reporting approximately 200 mostly isolated clinical cases have been published in the English literature. It has also been described with various terms such as medial cleft, median fissure of the neck, congenital midline cervical cord, midline cervical webbing, and pterygium colli

medianum (Erçöçen et al., 2002; Bajaj et al., 2004; Sinopidis et al., 2012). Until now CMCC has been a relatively unknown entity and there is much controversy over its etiology (Bajaj et al., 2004).

Thought it is always present at birth, it is often overlooked or misdiagnosed (Ayache et al., 1997). Inadequate treatment may cause secondary complications such as impaired neck extension, microgenia, exostosis, torcicollis, or infection (Sinopidis et al., 2012).

The purpose of this article is to present a case of a patient with CMCC and to do a brief review of the literature about the diagnosis, embryopathogenesis, and treatment of this rare entity.

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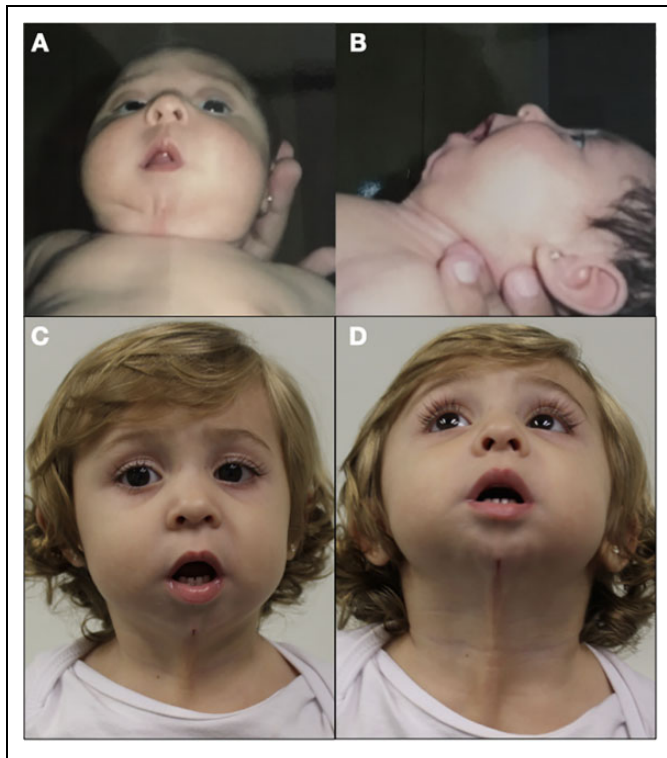Email: cristianotonello@usp.br

Figure 1. Clinical photographs of a 2-month-old girl illustrating mild micrognathia (A and B) and at 18-months old presenting a cutaneous retraction limiting full extension of the neck (C and D).

Case Report

A 2-month-old Caucasian girl was referred to the Hospital of Rehabilitation of Craniofacial Anomalies in Bauru, Brazil, presenting with micrognathia and a congenital anterior midline cervical anomaly.

Her mother was 27-years-old at conception. Both parents were healthy and nonconsanguineous and have kindly provided written informed consent for the case report to be published.

The pregnancy was uneventful but the patient was born after a cesarean delivery at 35 weeks of gestation due to a preterm rupture of membranes. The patient stayed in the intensive care unit during the first 5 days of life because she had respiratory distress caused by aspiration of secretions.

On examination, she had micrognathia, a notch in the mandibular symphysis and a region of cutaneous retraction with subcutaneous soft tissue thickening in the mandibular midline extending to the neck (Figure 1), without associated bone instability. The patient had no other known health problems.

Computer tomography showed abnormal tissue in relation to the anterior neck (Figure 2) and magnetic resonance imaging revealed a longitudinal linear lesion located in the subcutaneous fat plane of the midline anterior cervical area (Figure 3), extending between the chin and the thyroid isthmus. The lesion was 4.4 cm in craniocaudal, 1.6 cm in anteroposterior, and less than 1 cm in transversal dimensions, it presented an intermediate signal in T1- and T2-weighted sequences with noncontrast enhancement.
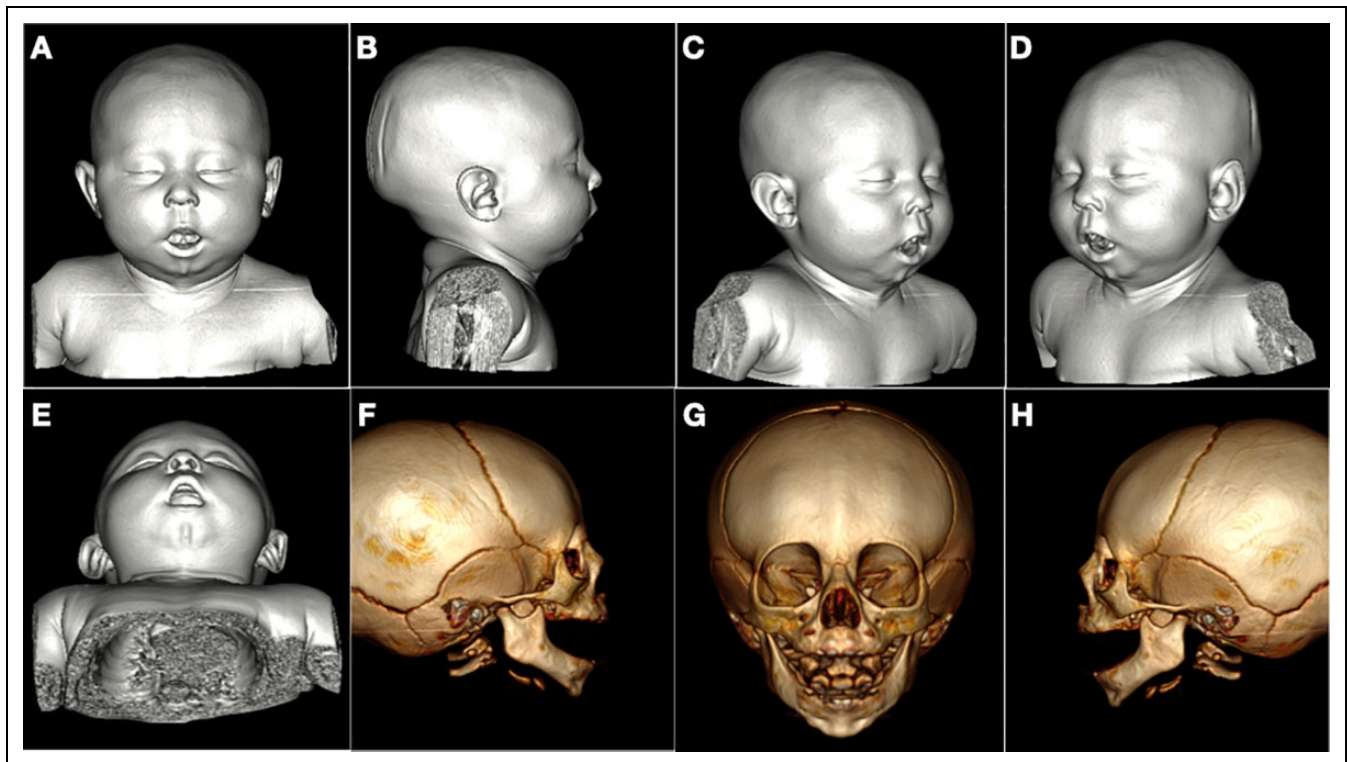

Figure 2. Three-dimensional computed tomographic (CT) images before surgery.
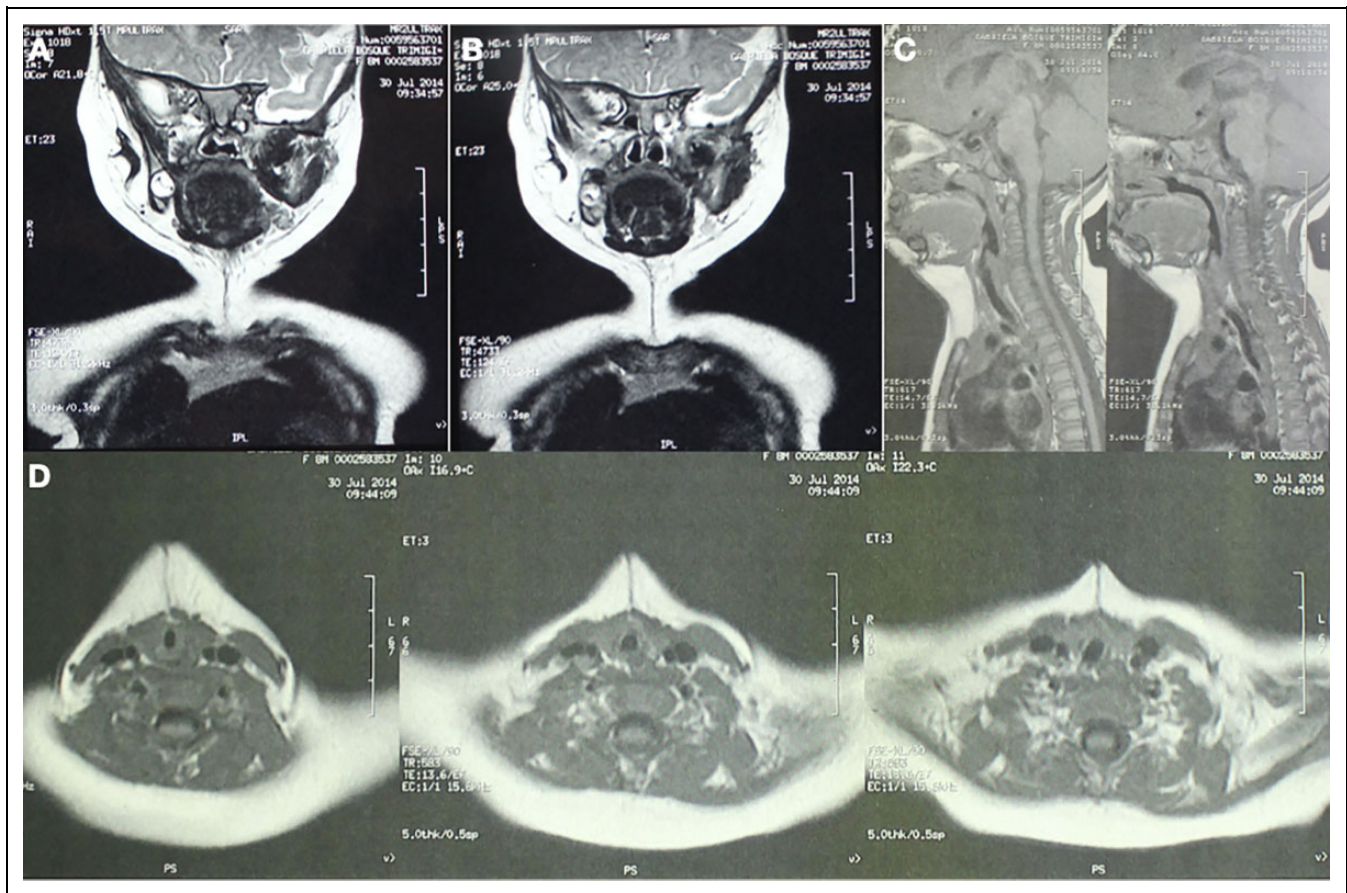
Figure 3. A 4.4-cm linear subcutaneous lesion of midline cervical area, extending between the chin and the thyroid's isthmus on magnetic resonance image.

The diagnosis of CMCC was made based on the clinical and radiologic features. At the age of 18 months, the patient was operated on under general anesthesia by the craniofacial surgery team, with excision of all the pathologic tissue and reconstruction with multiple Z-plasties (Figure 4).

Histopathology reported the presence of dermal fibrosis with a lymphocytic infiltrate. The patient was maintained under surveillance. The postoperative result at 3 years revealed stable micrognathia and minimal hypertrophic scarring (Figure 5). There was a good function result, with no cervical movement restriction.

Discussion

Characteristically, a CMCC consists of a vertical cutaneous and subcutaneous defect of variable length along the midline of the anterior neck, the majority of which are diagnosed at birth (Derbez et al., 2004; Cochran et al., 2006; Achard et al., 2016).

Several studies suggest there is a higher female prevalence with a ratio of 2:1 (Sinopidis et al., 2012; Eom et al., 2014; Goldfisher et al., 2015). Achard et al. (2016), however, found an equal ratio and Puscas (2015) even found a male predominance in his published retrospective series. Regarding

ethnicity, the actual literature describes a predominance in the Caucasian population (Bahakim et al., 2018).

No previous published article has suggested a familial inheritance pattern (McInnes et al., 2012). A genetic study, by Jakobsen et al. (2012), describes the potentially sporadic character of CMCC. He did a genetic analysis on 3 cases of isolated CMCC. Two mutations were found which can be contributing factors for this malformation: deletion of the pregnancy-associated plasma protein A and mutation in the SIX5 gene. Agag et al. (2007) found that CMCC was associated with 13/14 de novo Robertsonian translocations.

Congenital midline cervical cleft presents 3 specific clinical and pathological features:

1. A cranial nipple-like skin tag composed of stratified squamous epithelium and striated muscle (McInnes et al., 2012; Eom et al., 2014; Crippa et al., 2015; Puscas, 2015).
2. An erythematous, vertical, and atrophic skin defect in the midline of the neck comprised of stratified squamous epithelium without adnexal structures, with mild inflammation of lymphocytes, plasma cells, and neutrophils possibly present. A subcutaneous band-like fibrosis is present above the platysma plane which is often
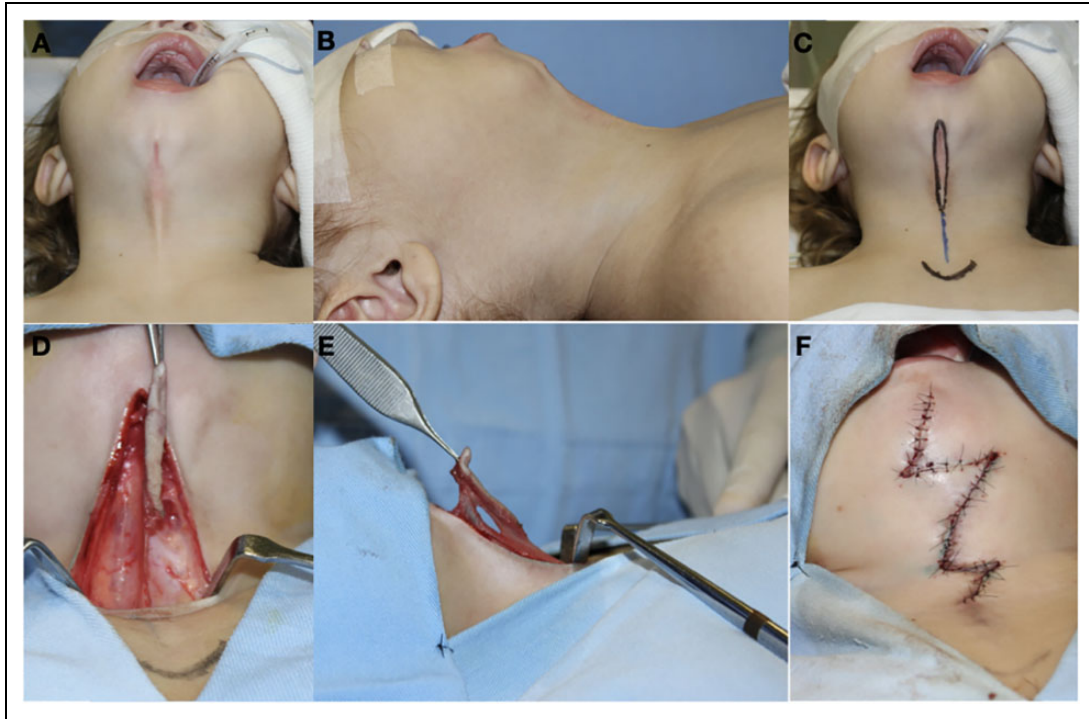
Figure 4. Preoperative image (A and B). Intraoperative resection of fibrotic subcutaneous cord (C-E) followed by a Z-plasty reconstruction (F).
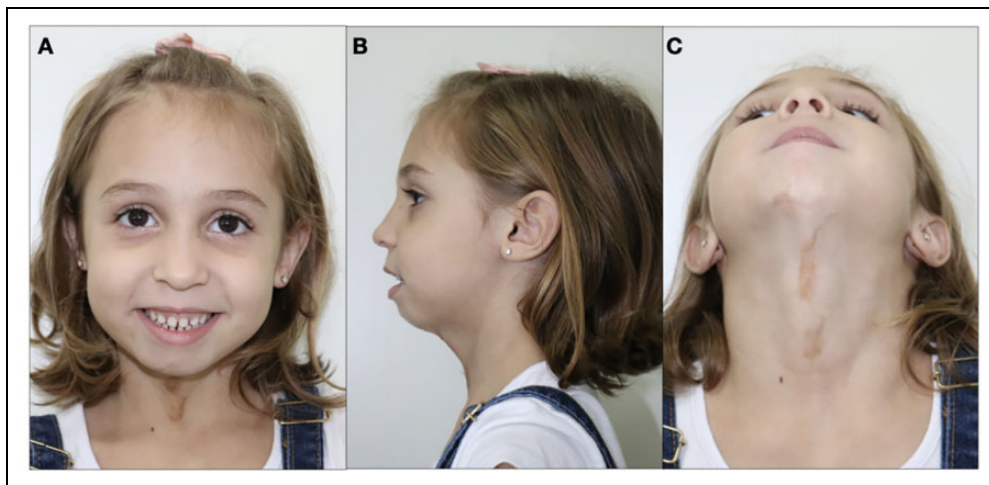

Figure 5. Three-year follow-up of surgical correction, revealing minimal hypertrophic scar and appropriate functional result.

longer than the overlying skin defect (Eom et al., 2014; Puscas, 2015; Bahakim et al., 2018).

3. A sinus tract at the caudal end composed of squamous or pseudostratified columnar epithelium which can discharge mucoid material. The source of the discharge is assumed to be from the presence of heterotopic salivary tissue inside the sinus tract which resolves gradually after a few months (van der Staak et al., 1991; Maschka et al., 1995; Erçöçen et al., 2002; Cochran et al., 2006; Sinopidis et al., 2012; Saha et al., 2013; Crippa et al., 2015).

The subcutaneous fibrous cord is always present and may represent the only feature in partial clinical manifestations (Crippa et al., 2015). It causes webbing in mild cases and flexion contracture in severe cases, limiting neck extension, or even depressing the lower lip during extension of the neck (Bajaj et al., 2004; Cochran et al., 2006; McInnes et al., 2012). Such a contracture is thought to be the result of healing of the cleft and tightening with subsequent growth and development (Erçöçen et al., 2002; Bajaj et al., 2004; Agag et al., 2007). This is because the affected tissues lag behind in vertical growth compared with the surrounding normal neck tissue (Puscas, 2015).

Other possible associated anomalies that can appear when the fibrous cord extends to the level of the mandible are micrognathia and presence of a mandibular bony spur secondary to the traction caused which may be severe enough to produce an open-bite deformity. That bony prominence can be palpated and/or visible on radiographic examination (Erçöçen et al., 2002; Bajaj et al., 2004, McInnes et al., 2012; Goldfisher et al., 2015).

The embryopathogenesis of this anomaly has not been clearly established (Achard et al., 2016). It is considered part of the midline branchiogenic syndromes, but it is not a true cleft in the same way as a cleft palate. Many authors have concluded that incomplete fusion of the second branchial arches is largely responsible for this entity, and several mechanisms have been proposed to explain it: the presence of amniotic adhesions and vascular anomalies may cause localized tissue ischemia, necrosis and scarring of the developing branchial arches, or pressure on the developing cervical area by the closely juxtaposed pericardial roof during the fifth week of gestation may produce similar results. A disturbance in the interaction between the mesoderm and the ectoderm may explain the lack of adnexal elements in the skin defect (Stephen et al., 1989; Puscas, 2015; Bahakim et al., 2018).

The extent of mesodermal proliferation within the cleft to close the fusion's gap determines lesion severity. Gargan et al. (1985) hypothesized that CMCC can fall into 2 main groups: (1) isolated when there is decreased cellular migration through the second arch, and (2) extensive when there is incomplete fusion of the second arch derivatives with the first and/or third arch (Saha et al., 2013; Bahakim et al., 2018). In consequence, the CMCC has been associated with a spectrum of midline anomalies related to the branchial arches, including cleft of the lower lip, mandible, tongue, sternum, hypoplasia, or aplasia of the hyoid bone or thyroid cartilage (Derbez et al., 2004; McInnes et al., 2012; Sinopidis et al., 2012).

Other possible associated anomalies are congenital cardiac defects and thyroglossal, bronchogenic, or dermoid cysts (Ayache et al., 1997; Bajaj et al., 2004; Crippa et al., 2015).

Although the diagnosis is made on clinical presentation, imaging may be requested to the differential diagnosis or to determine any coexisting lesions. Magnetic resonance imaging is the best modality to determine the extent of the tract, associated ENT anomalies, and for presurgical planning (Goldfisher et al., 2015).

To avoid complications of long-standing CMCC early intervention is recommended; however, no general consensus exists as to an appropriate age for treatment (Crippa et al., 2015).

Surgery is established as being the reference treatment for CMCC which consists in complete excision of the lesion followed by closure of the defect (McInnes et al., 2012; Goldfisher et al., 2015; Achard et al., 2016). Simply transecting the fibrous cord or performing incomplete excision of the cutaneous and subcutaneous elements leads to recurrence (Bajaj et al., 2004; Puscas, 2015).

Several methods of surgical correction of CMCC defects have been described including linear closure and single or multiple Z-plasties or W-plasties (Ayache et al., 1997; Bahakim et al., 2018). The success is highly variable depending on the location, size, and length of time before surgical correction and the complete excision of the lesion (McInnes et al., 2012).

Linear closure has been suggested as a possible option for small lesions and the surrounding skin is lax. However, when used in long defects, it can result in hypertrophic scarring, contractures, and secondary anterior open bite (Cochran et al., 2006; Puscas, 2015).

The use of single or multiple Z- or W-plasties is recommended for longer defects to improve the cosmetic and functional results and have less risk of cicatricial contracture formation (Bahakim et al., 2018).

Z-plasty is the most commonly used way of incision closure in the literature. It allows the surgeon to (1) lengthen a contracted scar, (2) reorient the direction of a scar or defect, (3) breakup a straight line, and (4) shift soft tissue contour. Despite that Z-plasty can lead to hypertrophic scarring in the oblique limbs along with triangle tip depression and necrosis if the angles are too acute (<30°). For that reason, the result of the operation is not always regarded as being cosmetically successful by the parents. In general, however, the result is functionally good and the patient is able to extend his neck without any impediment (van der Staak et al., 1991; Crippa et al., 2015; Bahakim et al., 2018).

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