CASE REPORTS

A case of untreated midline cervical cleft associated with congenital heart defect

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ABSTRACT

A congenital midline cervical cleft (CMCC) is a rare anomaly. Less than 100 cases report in English literature have been reported. This failure is presented at birth and in most cases is treated in the first months of life. We report an unusual case of a 14-year-old girl with untreated CMCC, which was misdiagnosed as thyroglossal duct cyst. The patient also presented rare congenital heart disease. In this case report we discuss the clinical presentation, diagnosis and surgical treatment. The aim of this case report is to draw attention to very rare case of patient with misdiagnosed CMCC.

Key Words: Congenital midline cervical cleft, Branchial cleft, Congenital heart disease, Z-plasty

1. INTRODUCTION

Congenital midline cervical cleft (CMCC) is a rare anomaly of anterior neck and very often followed by misdiagnosis. Less than 100 cases report in English literature have been reported. The origin of CMCC is not clearly resolved. It is assumed of impaired fusion of 1^{st} and 2^{nd} branchial arches as the most acceptable explaining in 3^{th}-4^{th} week of embryogenesis.[1] Bailey made the first description in 1924.[2] The lesion varies includes nipple-like skin appendix in the superior portion, cleft in the middle portion and sinus tract in the inferior portion,[3] aslo there can be lack of hyoid or thyroid cartilage. It can be associated with cleft of lower lip or cleft tongue and rarely with a bronchogenic cyst or with respiratory epithelium, hemangiomata.[4] Associated congenital heart defects have been reported occasionally, although most of these cases have been associated with cleft sternum[4] or ectopia with intracardiac anomalies.[5,6]

2. CASE REPORT

We present case report of a 14-year-old girl with CMCC associated with congenital heart lesion. Girl had midline skin lesion in the anterior part of the neck since birth misdiagnosed as thyroglossal duct cyst. In her medical history there was cyanotic single ventricle with pulmonary valve stenosis and malposition of the great blood vessels treated surgically after birth.

Neck anomaly had all three signs of CMCC, including nipple-like skin appendix, midline cleft and caudal sinus with associated submucous fibrous cord. This palpable fibrotic cord was running subcutaneously towards sternum in midline platysma 8 cm long and 1 cm weigh and performed restriction in dorsal flexion of the head and neck (see Figure 1).

There was positive history of inflammation of the blind ending sinus with purulent discharge and swelling. Examination of the rest of the face, neck and oral cavity was normal; there

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was no other sinus or cleft.

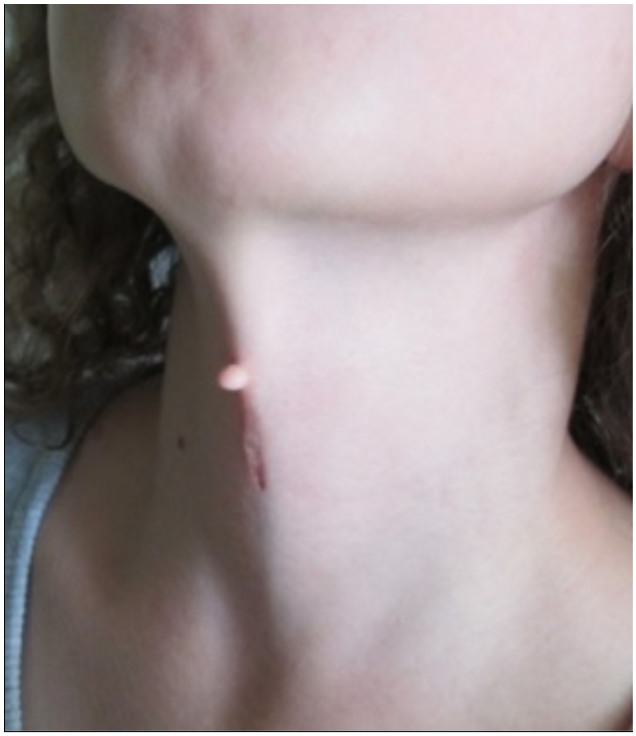

Figure 1. A 14-year-old girl with CMCC before surgery

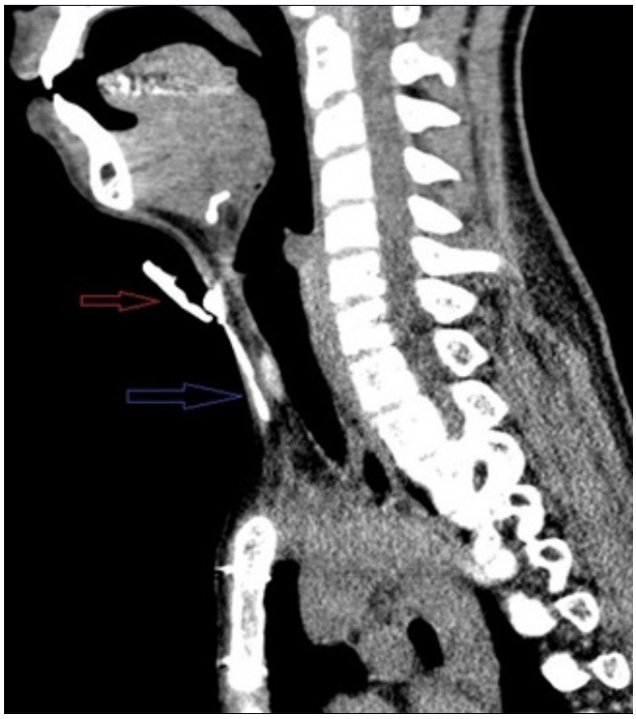

Figure 2. CT with blind ended sinus filled with contrast agents and fibrotic restricting cord
Red arrow (intravenous cannula used for contrast instillation); Blue arrow (blind ended fistula filed with contrast agents)

After unclear ultrasonography result CT scan was performed, instilling contrast agents it to the sinus with venous flexible cannula (see Figure 2). CT scan cleared the diagnosis of CMCC by imaging distal blind ended fistula, fibrotic restricting chord and nipple like skin lesion. This figure haven't been published yet.

Surgery under general anaesthesia was performed and complete excision of the cleft with underlying fibrotic tissue was made and the vertical wound was closed by primary Z-plasty (see Figure 3). There were no post-operative complications and the scar was healing very well at 10^{th} day follow up (see Figure 4).

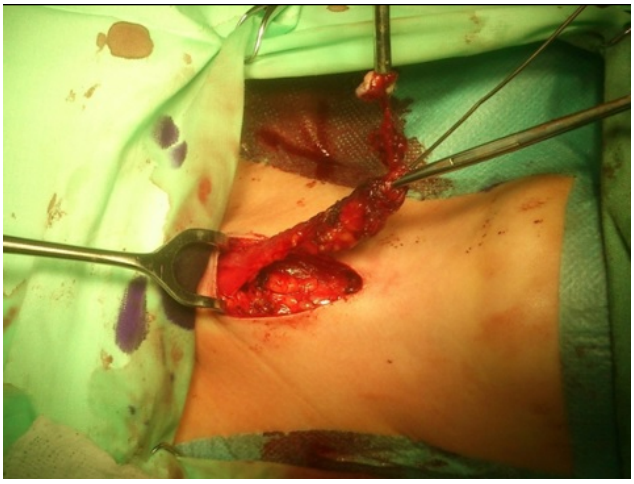

Figure 3. Complete excision of the congenital midline cervical cleft under general anaesthesia

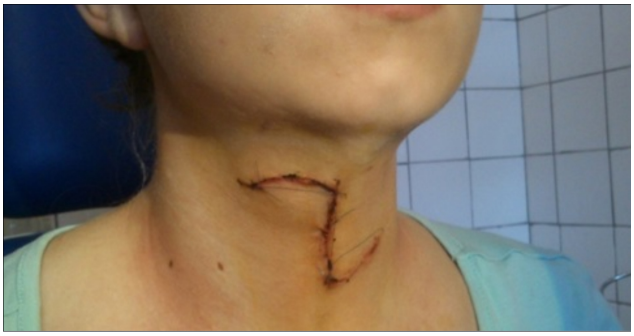

Figure 4. The scar healing at 10th day follow up

On histology, the nipple-like structure consisted of acantotic epidermis with skin appendages in dermis. The cleft portion was covered by parakeratotic epidermis and structures with signs of branchial cyst cylindrical epithelium. The sinus tract was lined by pseudostratified ciliated columnar epithelium associated with rare seromucinous glands and chronic inflammation (see Figure 5). Histological findings were consistent with the diagnosis of CMCC.

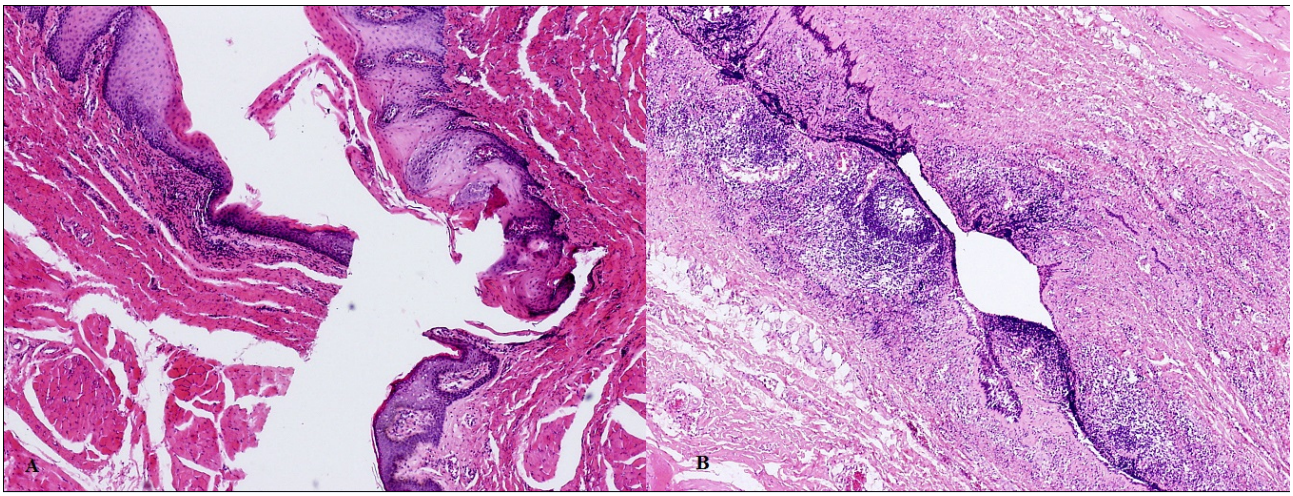

Figure 5. A histopathological figure of CMCC 40× optical zoom
A. External orifice of skin without adnex; B. Caudal end of fistula

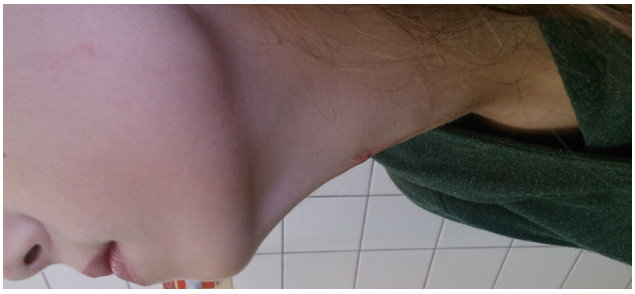

Figure 6. Follow up after two years after surgery. Neck contraction in dorsal flexion of the head and neck.

A follow-up examination carried out two years after the surgery revealed good wound healing of the Z-plasty, no history of discharge. However, neck contraction was presented again in dorsal flexion of the head and neck (see Figure 6). The patient has been therefore scheduled for reoperation.

3. Discussion

CMCC is rare congenital malformation which can be associated with other congenital lesions. It is manifested after birth with three typical signs.

Hirowaka *et al.* reported CMCC case with atrial septal defect lesion, however most of these cases have been associated with cleft sternum or ectopia with intracardiac anomalies.[6] We present other CMCC case associated with cardiac lesion as a possible proof of Hirowaka hypothesis about broad spectrum of CMCC associations with cardiac lesions. Hirokawa reported partial septal defect and our patient had ventricle defect with pulmonary valve stenosis and malposition of the great blood vessels.[6]

Although we present CT scan with complete imaging of typical distal blind ended fistula filled with contrast agents which haven't been presented yet.

Untreated CMCC may cause neck contractures and deformations of mandibula and sternum. The purpose of early surgery is to avoid cicatrical contractures and mandibular deformities such as exostosis-like protuberances, both of which are more likely to occur as the child gets older.[4,7,8]

Many authors suggest that a proper time for the surgery in children is before the second birthday, with earlier repair being indicated in more severe cases. Delayed surgical treatment of such congenital anomaly can lead to recurrence of neck contracture as described in our case report. Therefore, an early resolution of this disorder is important for the successful outcome of the surgical treatment.

Surgical golden standard is complete excision of pathological tissue with skin, platysma, fibrotic cord and the multiple or simple Z-plasty, which are believed to help avoid cicatrical contractures and vertical scar.[8,10,11] However the severity of regional hypoplasia decides the reconstructive armamentarium ranging from Z and V-Y plasty for simple webs to tissue expansion or myocutaneous flaps for severe regional hypoplasia and geniosternoplasty for mentosternal clefts.[12,13]

Simple closures are not cosmetically pleasing in the long run due to recurrent scarring. Straight line closure leads also to a higher incidence of neck contracture recurrences. Therefore it is recommended to close the defect using multiple Z-plasty technique.

There are reports from various specialists in literature – plastics, reconstructive, ENT and dermatologic depends on primal misdiagnosis. A 14-year-old girl was send thyroglossal duct cyst to pediatric ENT department. It is rare lesion and

there is importance of correct recognition and appropriate surgical management.

Table 1. Differential diagnosis of the CMCC

• Thyroglossal duct cyst
• Thyroglossal fistula
• Branchial cleft cyst
• Dermoid cyst
• Epidermal inclusion cyst
• Fibroepithelial polyp
• Pilomatrixoma in the midline of the neck
• Bronchogenic cyst

The differential diagnosis includes many congenital anomalies (see Table 1), especially branchial, thyroglossal and chondrobranchial remmants. Although CMCC is easily differentiated by its typical presentation, imaging studies are needed (CT scan, MRI).[14] Most often it is mistaken for a thyroglossal duct cyst or fistula. These midline T2 hyperintense lesion may have faint peripheral enhancement, characteristically appears cystic and it is typically much higher in the neck than CMCC.[15] In addition, a thyroglossal duct cyst does not usually have an associated sinus tract unless it is superinfected. 2^{nd} or 3^{rd} branchial cleft cysts or pyriform sinus fistulae are typically off midline in the lateral neck.[15]

4. Conclusion

The CMCC is a rare anomaly in which an early diagnosis is crucial and should not be omitted. Although the suitable time of CMCC surgery is not clearly mentioned, based on our result the earliest possible time of intervention is recommended for successful result of the operation. Untreated CMCC results in severe neck contracture and following surgical treatment could have less positive outcome.

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