Late Presentation of a Congenital Midline Cervical Cleft Involving the Thyroid: An Uncommon Anomaly

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Introduction

The congenital midline cervical cleft (CMCC) was first described by Bailey in 1924.1 Although variations in presentation exist, CMCCs are typically seen as a vertical midline ventral cervical defect with a cranial skin tag with a short mucosalized sinus and varying degrees of subcutaneous fibrous banding. Fewer than 100 cases have been reported, and its etiology remains unclear.2 Multiple theories have been proposed, most of which include defects in anterior fusion of the first and second branchial arches.3

Other hypotheses have been proposed. Adhesions may form between the developing heart and the branchial arches, causing defective fusion between them.4 Others have proposed that first branchial arch mesoderm develops an abnormal additional growth.5 A persistent midline sulcus in the first branchial arch related to incomplete differentiation of the mesoderm may impair the fusion of more caudal arches.5

This anomaly appears to be more common in females, and treatment universally involves surgical resection of the lesion in infancy for cosmesis, to avoid cervical contracture and limitation of extension, and to avoid episodes of recurrent drainage.6 There is no known genetic cause of or association with CMCC; however, several candidate genes have been identified.7

Case Report

We present an 8-year-old female who was referred for evaluation of a congenital anterior neck lesion, which was disfiguring and draining chronically. The patient was born full-term and the perinatal period was uneventful. There was no family history of similar lesions.

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Pathology

Pathology showed skin with underlying dermal scar and mild chronic inflammation in the superficial dermis.

Imaging

A CT scan had been performed and revealed a band of muscular density tissue extending inferiorly from the inferior border of the mandible, immediately below the level of the skin for approximately 7 cm in the midline to the suprasternal region. There was a posterior extension of fat-density splitting the thyroid gland into two without obvious connection to the trachea. A single midline vessel was noted within the lesion.

Figures

Figure 1 – A representative image of a CMCC (taken from Warden & Millar).8

Discussion

The CMCC is a rare malformation with variable presentation and is not only aesthetically displeasing. The typical description of CMCC is a midline anterior neck defect with a craniocaudal orientation, a nipple-like skin tag cranially with a sinus tract caudally.9-10 Neck movements may be restricted.11-12 Defects can be severe as in a reported case of a patient with CMCC and a cleft of the lip, mandible and sternum, as well as absence of the hyoid bone and thyroid cartilage.12 One patient had a CMCC associated with a bifid tongue, floor of mouth and hypoplastic strap muscles.2

The tissue types involved in these lesions may vary. The skin tag appears as a bulge with normal overlying skin, but may contain other tissues such as cartilage or skeletal muscle.13 The cleft is generally lined by parakeratotic stratified squamous epithelium (with or without skin appendages) and the dense fibrous tissue underlying the dermis may have an inflammatory infiltrate or adipose tissue.13 There is typically a mucosalized tract with cylindrical epithelium.7

Treatment universally involves surgical resection of the cleft and it appears important that all pathologic tissue be removed. In one report, the skin tag and caudal sinus tract were excised with two small transverse ellipsoidal incisions, and the fibrous band was removed subcutaneously in an attempt to avoid a larger vertical incision.6 In this case, the fibrous cord recurred causing webbing and contracture, ultimately requiring reoperation.1 However, there is a report of a patient with an atypical variant of a CMCC who lacked the typical caudal sinus tract and mucosalized tract was treated successfully with excision of the skin tag and dissection of the underlying fibrous cord.14

The preferred closure is with Z-plasties to minimize the appearance of the scar and decrease webbing, however straight closure has been used with variable results.12,13,15-17 In 3 cases, healing after linear closure was uneventful and yielded acceptable cosmetic results.15,16,18 In other reports patients developed contractures, hypertrophic scars, and in one case an anterior open-bite deformity.12,15 Gargan et al. noted that although the horizontal scars of the Z-plasty were minimal, the oblique scars may become hypertrophic.15 Others reported superior tethering and cosmetically unsatisfactory results.3,16 In many reports Z-plasty is utilized with good results and most authors consider it to be the method of choice for these patients, however the length of follow-up in these reports is variable.15,19,20

Conclusions

Midline cervical clefts are an uncommon congenital anomaly without a well-described etiology. Diagnosis is based on clinical exam of a midline cervical lesion with a superior skin bulge and associated mucosalized sinus and underlying fibrous band. Imaging is used as an adjunct to delineate the extent of the cleft including involvement of other structures. Surgical excision is curative and early intervention may decrease the chance of contracture and deformity.

References


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These images are NOT of the authors’ patient, but are representative of CMCC.