Congenital Midline Cervical Cleft

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Overview

Background

The congenital midline cervical cleft is a rare developmental abnormality of the anterior part of the neck Luschka in 1848 and noted in the English literature by Bailey in 1924 [1,2]. The lesion is often initially evaluated by a pediatrician or primary care physician who may misinterpret the anomaly as a branchial cleft deformity or thyroglossal duct cyst. Although developmentally related to these disorders, the congenital midline cervical cleft represents a distinct anomaly that should be recognized at initial examination. The parents can then be appropriately counseled about the implications and further management decisions that are peculiar to this disorder.

Pathophysiology

The embryologic mechanism that accounts for the congenital midline cervical cleft is not firmly established; however, most investigators believe that it involves a failure of the branchial arches to fuse in the midline [3,4]. In normal embryology, the branchial arches grow medially and merge in a cephalad to caudal direction with the first (mandibular) arch closing before the second (hyoid) arch, and the lower arches following sequentially. Prior to fusing, mesodermal tissue migrates between the arches, pushing ectoderm outward to flatten the ventral furrow.

A number of hypotheses have been set forth to explain the faulty closure of the branchial arches. For instance, mechanical factors and vascular anomalies with secondary ischemia and necrosis have been postulated in the mechanism [5]. Failure of the mesenchyme to penetrate the midline, with subsequent poor interaction between the mesoderm and ectoderm, has also been suggested as a means of preventing fusion [2,6-9]. This mechanism could also explain the absence of normal adnexal structures that is observed in involved skin [10].

Finally, a few investigators postulate that pressure on the cervical area by the pericardial roof could prevent fusion and another takes the debate to a microscopic level with proposed roles for hyaluronate and collagen [3,9-11].

Epidemiology

Frequency

The incidence of congenital midline cervical cleft is difficult to evaluate. In 1985, Gargan et al. reported 12 cases of midline cervical clefts as part of their 30-year series of 612 thyroglossal and other branchial cleft sinuses [11]. This represented an incidence of 1.7% in this family of disorders. In a similar series of 198 cases by Gross and Connerly in 1940, only two defects were identified as being midline clefts [12]. In a more recent series of 191 congenital cervical malformations by Nicollas and Guelfucci in 2000, an incidence of 2% midline cervical clefts was reported.

Mortality and Morbidity

Many congenital midline cervical clefts are asymptomatic. There always exists a subcutaneous cord-like, fibrous thickening cephalad to the defect, which may cause webbing. In severe cases this fibroid cord may form a cervical contracture of the anterior neck that becomes more pronounced as the patient matures [13,14].

- Race: Unknown.
- Sex: Unknown.
- Age: Midline cervical clefts are rare anomalies that are present at birth but may be overlooked or misdiagnosed. It usually becomes more obvious with time.

Presentation

History

Congenital midline cervical cleft is clinically typical and diagnosed at birth. Length and width can differ from one child to another but clinically cleft presents as linear vertical areas of thin erythematous skin. Sometimes mistaken by the patient’s family as a “birthmark,” some patients may also present with a working diagnosis consisting of thyroglossal duct, dermoid, or branchial cleft cyst.

Physical

The lesion presents at birth with a ventral midline defect of the skin of the neck. A reddened weeping strip of atrophic skin approximately 5mm in width may occur at any level between the chin and sternal notch. Often there is a nipple-like projection at the upper end of the fissure and an associated sinus tract at the caudal aspect, which may discharge mucoid material. There always exists a subcutaneous cord-like, fibrous thickening cephalad to the defect which may cause webbing, also known as pterygium colli medium. In severe cases this fibroid cord may form a cervical contracture of the anterior neck that becomes more striking as the patient grows [15].

In addition to its central features, congenital midline cervical cleft may be associated with other anomalies of the head and neck, including clefts of the lower lip, mandible, chin, and tongue, an underdeveloped mandible, and hypoplasia/absence of supporting structures of the neck, such as the hyoid bone. It has occurred in the presence of, and may even be related to, both bronchogenic cysts and thyroglossal duct abnormalities. Furthermore, congenital midline cervical cleft has been associated with defects in other parts of the body, including cleft sternum, midline abdominal web or scar-like raphe, and midline hemangioma, as well as congenital heart lesions such as ectopia and intracardiac anomalies.

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Causes

Developmental anomalies of branchial arches can provoke several malformations. The main theory hypothesized that midline clefts are the consequence of an anterior fusion defect of the two first pairs of branchial arches during the third and fourth weeks of intrauterine life [3].

Workup

Laboratory studies

No specific laboratory studies need to be obtained in the workup of a congenital midline cervical cleft.

Imaging studies

A sinogram may be obtained. If a sinus tract exists, radioopaque dye can be injected to delineate the course and to examine the size of the cyst.

Ultrasonography can be helpful especially if a thyroglossal duct cyst is suspected. Thyroid aspect and position are also specified [16].

A contrast-enhanced CT or MRI scan is not contributive.

Other tests

No other investigations are needed beyond routine preoperative workup.

Procedures

Fine-needle aspiration has been reported to be helpful in distinguishing branchial cleft cysts from malignant neck masses. Fine-needle aspiration and culture may help guide antibiotic therapy for infected cysts.

Histologic findings

The histology of the congenital midline cervical cleft usually consists of parakeratotic stratified squamous epithelium without normal skin appendages [10,15,17]. The underlying dermis may or may not show areas of alternating hypertrophy and atrophy, and the subcutis may contain dense fibrous connective tissue and a mild inflammatory infiltrate of primarily lymphocytes, plasma cells, and neutrophils [10]. The skin tag generally consists of normal overlying skin, but can contain cartilage and irregular skeletal muscle [17]. The sinus tract associated with the congenital midline cervical cleft usually consists of pseudostratified columnar or cylindrical epithelium and often demonstrates adjacent seromucinous glands [10,18]. In some cases, however, the histology of this fistula resembles bronchogenic epithelium or contains part of the thyroglossal duct or its remnants [2,7,19]. Finally, the fibrous cord that overlies the cervical midline cleft has its own distinct microscopic structure, and may include interfasciculated bundles of skeletal muscle that resemble torticollis histologically [8].

Treatment

Medical care

Antibiotics are required to treat infections or abscesses related to congenital midline cervical clefts.

Surgical care

The recognition and early surgical intervention of congenital midline cervical clefts have important functional and cosmetic implications [6]. Failure to recognize the stigmata of this abnormality and delay of surgical treatment can lead to cicatricial contracture and potentially mandibular hypoplasia. No general consensus exists as to an appropriate age for elective surgical resection of midline cervical clefts.

Several methods of surgical correction of have been described, yet most authors would recommend Z-plasty technique to correct anterior cervical contracture and obtain optimum cosmetic result [9-12]. However, the cases reported in the literature generally represent the most florid examples of midline cleft. If the defect is small and adjacent tissues are lax and the fibrous cord is completely excised, the defect can be closed primarily [15].

Z-plasty is a common technique and a versatile surgical maneuver. Z-plasty allows the surgeon to (1) lengthen a contracted scar; (2) reorient the direction of a scar or defect; (3) break up a straight line; and (4) shift soft tissue contour [20,21]. The Z-plasty technique involves creating two opposing triangular transposition flaps that are rotated synchronously to close a central defect by redistribution and rearrangement of tissue [22]. The Z-plasty is symmetrically designed so that the lateral limbs are equal in length to the tissue defect (central limb) and that the angles between the lateral limbs and the central limb are 60 degrees. Re-approximation of the central defect at the skin level prior to designing the Z-plasty flaps has been noted to aid in constructing a more precise Z-plasty [23,24].

The angle of the Z-plasty flaps may vary, and the gain in length varies directly with the angle of the Z-plasty. The optimal angle has been determined to be 60 degrees, which has a theoretical gain in length of 75%. The angle may be greater than 60 degrees, but the tension required to transpose skin flaps increases as the angle of the Z-plasty increases such that angles greater than 75 degrees cause tissue distortion and dog-ear deformities. Angles less than 20 degrees present problems with flap viability secondary to compromised blood flow at the flap tips [25].

Long-term functional results of the congenital midline cervical cleft Z-plasty closure have been reported to be satisfactory with improvement in vertical neck movement. However, cosmetic results are more variable. Horizontal scars often remain thin but widening of the oblique limbs of the Z-plasty scar has been described [9,10].

Consultations

Referral to an Otolaryngologist for surgical excision is indicated.

Follow up

Further outpatient care

Postoperatively, patients should be monitored for appropriate healing after excision.

Complications

Untreated congenital midline cervical clefts cause cicatrical contracture and changes in the mandible, which include submental bony prominence, micrognathia, mandibular hypoplasia and mild cleft mandible all thought to be secondary to traction on growing mandibular bone.

Complications of surgical excision of congenital midline cervical
Congenital midline cervical clefts result from inadequate removal, damage to surrounding blood vessels and nerves, and inadequate closure leading to scar formation.

**Prognosis**

Following surgical excision of congenital midline cervical clefts, recurrence is uncommon.

**Patient education**

Patients should be reassured that congenital midline cervical clefts are benign.

**References**