Congenital midline cervical cleft: A report of 3 cases

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Abstract

Congenital midline cervical cleft is a rare anomaly of the anterior neck. The diagnosis is typically made on the basis of the lesion’s characteristic clinical presentation at birth. It appears to occur as a result of a failure of fusion of the paired second branchial arches in the midline during embryogenesis. Surgical intervention is necessary to avoid potential long-term complications, such as contractures and limitation of neck mobility. We present 3 cases of congenital midline cervical cleft, and we discuss the embryology, presentation, and surgical management of this unusual condition.

Introduction

Congenital midline cervical cleft is an unusual condition identified at birth. Clinically, the presenting signs appear along a wide spectrum, ranging from a simple cleft of the soft tissue to an extreme defect associated with clefting of the mandible and/or sternum and a possible loss of other midline structures, such as portions of the hyoid bone. In most cases, however, the lesion presents as a vertically oriented cleft of pink tissue in the anterior midline neck, often with an associated protuberance of skin superiorly and a blind sinus tract inferiorly. We report our management of 3 patients with this lesion.

Case reports

Patient 1. Following an uncomplicated pregnancy and delivery, a full-term newborn girl was referred for evaluation of a congenital anterior neck lesion. The patient had no airway or feeding difficulties, and her cry was normal. Physical examination revealed a pink, moist region in the ventral midline neck. A protuberance of skin was noted superiorly, and a short sinus tract was noted inferiorly; a subtle notching of the mandible was also observed.

Excision of the lesion was deferred until the patient reached the age of 12 months. At that time, she underwent a complete excision of the inferior sinus tract and superior protuberance (figure 1, A). Closure was performed with multiple Z-plasties (figure 1, B). Pathology of the skin identified acanthosis and parakeratosis. Also present were mucinous salivary glands. At follow-up 2 years later, no contracture was evident and the cosmetic result was excellent.

Patient 2. A full-term girl born during an uncomplicated delivery was noted to have a midline neck abnormality—specifically, a 5-mm protuberance of skin and a layer of subcutaneous tissue over the cricoid cartilage. Inferior to the abnormality was a vertically oriented strip of pink tissue that extended 2 cm to a blind sinus tract. The tract was probed to a depth of 4 mm. The platysma was tethered, and the mentum was slightly notched. A barium swallow test and a fistulogram confirmed the presence of the blind pouch. Computed tomography (CT) detected no other abnormalities. A diagnosis of midline cervical cleft was made.

When the patient had reached 11 months of age, she underwent surgical excision, release of the platysmal banding, and Z-plasty closure. Pathology identified a benign squamous epithelium-lined cutaneous sinus (figure 2). The patient did well postoperatively, and the cosmetic result was good.

Patient 3. A 6-month-old boy was referred for evaluation of stridor and a midline neck lesion that had been present since birth. Physical examination revealed a pink, moist region in the ventral midline neck. A protuberance of skin was noted superiorly, and a short sinus tract was noted inferiorly; a subtle notching of the mandible was also observed.

Excision of the lesion was deferred until the patient reached the age of 18 months. At that time, he underwent a complete excision of the inferior sinus tract and superior protuberance (figure 1, A). Closure was performed with multiple Z-plasties (figure 1, B). Pathology of the skin identified acanthosis and parakeratosis. Also present were mucinous salivary glands. At follow-up 2 years later, no contracture was evident and the cosmetic result was excellent.

A preoperative modified barium swallow examination and CT of the head and neck were scheduled, but the patient did not return for treatment for 1 year. When he did return, at the age of 18 months, his stridor had resolved, and a repeat fiberoptic laryngoscopy revealed that his vocal fold mobility was normal bilaterally. The lesion was excised...
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Figure 1. Patient 1. A: Preoperative photograph shows the deformity 12 months after the patient was born. B: Following excision, the defect is closed with multiple Z-plasties.

Figure 2. Patient 2. Postoperative photograph shows the excised benign squamous epithelium-lined cutaneous sinus.

and closed with a double Z-plasty. Pathology identified epidermal hyperplasia and parakeratosis without skin appendages. Seromucinous salivary tissue and scattered bundles of smooth muscle were present. The patient was lost to clinic follow-up, but a telephone call to his mother indicated that the cosmetic result was satisfactory.

Discussion
Only 50 cases of congenital midline cervical cleft have been described in the English-language literature. Eastlack et al reported a female-to-male ratio of approximately 2:1. Cases appear to be sporadic, as no reports have suggested a familial inheritance pattern.

The literature reflects a spectrum of clinical findings. The most consistently reported features are a vertically oriented cleft of reddened tissue, a protuberance of skin superiorly, a blind epithelium-lined sinus tract caudally, and a fibrous subcutaneous cord. Mucoid material may be expressed from the orifice of the sinus. Findings are, however, quite variable, occasionally involving only midline webbing without skin lesions. The cord may limit neck movement, and one or two bony prominences are frequently noted at the inferior aspect of the mandible; these bony protuberances seem to be secondary to traction from the cord. These features tend to become more prominent with time, making early intervention appropriate. Classically, there is some degree of retrognathia, and various degrees of clefting of the lip and mandible may be present. Clefting may extend inferiorly to involve the sternum and superiorly to the hyoid. Other anomalies that have been found in association with midline cervical cleft are thyroglossal duct cysts and ectopic bronchogenic cysts. Histology of the cleft will reveal atrophic skin with parakeratosis in the absence of adnexal structures. Salivary gland tissue is frequently found, as are patterns suggesting bronchial epithelium, which has led some authors to propose that the lesion has a respiratory origin. The sinus tract is typically lined with benign squamous or pseudostratified columnar epithelium. The subcutaneous cord has been shown to contain connective tissue and muscular bundles similar to findings in congenital torticollis.

Suggestions of a thyroglossal duct or bronchogenic origin notwithstanding, the consensus is that the lesion is caused by a failure of the fusion of the paired second branchial arches in the midline during the third and fourth weeks of fetal development. This associates the cervical cleft with other midline clefts such as cleft lip and cleft palate, which are caused by a failure of fusion of the first branchial arch. Various mechanisms of improper fusion have been proposed, including (1) ischemia and necrosis secondary to mechanical factors and vascular anomalies, (2) failure of the mesenchyme to penetrate the midline, resulting in poor interaction between the mesoderm and ectoderm, and (3) pressure exerted on the cervical area by the pericardial roof.

The diagnosis is easily made by simply observing the characteristic clinical findings present at birth. Some authors...