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IMAGES IN CLINICAL PRACTICE

CONGENITAL STERNAL CLEFT - ISOLATED OR A POSSIBLE PHACES SYNDROME?

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The authors describe a case of a female full-term baby born by vaginal delivery with Apgar scores of 9 at 1 minute and 10 at 5 minutes. The immediate physical examination revealed a skin defect from the xiphoid process, which was visible through the superficial fascia, that extended to the umbilical cord (Figure 1). The pregnancy was monitored and uneventful and there was no prenatal diagnosis. A 3 cm wide hemangioma was noted on the dorsal region and a discontinuity of the right collar bone was palpable. The baby was admitted to the intensive care unit for surveillance and evaluation. The chest and abdominal radiographs did not reveal any other malformations. Echocardiogram showed a normal cardiac structure with a small restrictive patent ductus arteriosus and a patent foramen ovale. Thoracic computed tomography confirmed a bifidity of the manubrium (maximum distance of 4,1 mm, Figure 2), a medial cleft from the sternal body to the xiphoid process and a median defect of the abdominal wall that extended to the umbilical cord. Abdominal ultrasound confirmed supraumbilical abdominal rectus diastasis with a maximum interrectus distance of 11 mm. By the 10th day of life there was a partial epithelization of the wound (Figure 3) and she was discharged home, without any other complication, with a complete healing of the skin defect observed two months later.

What is the most likely diagnosis?

A congenital sternal cleft results from an embryological defect in the fusion process of the two sternal bars. It corresponds to 0.15% of all the chest wall malformations and seems to be more common in females.[1,2,3] Although it is a rare condition, PHACE syndrome must be considered when assessing a sternal cleft. It is an acronym for posterior fossa anomalies, hemangioma, arterial anomalies, cardiac anomalies, and eye anomalies.[4] In the presence of ventral developmental defects, such as sternal cleft or supraumbilical raphe, the term "PHACES" is used.[5]

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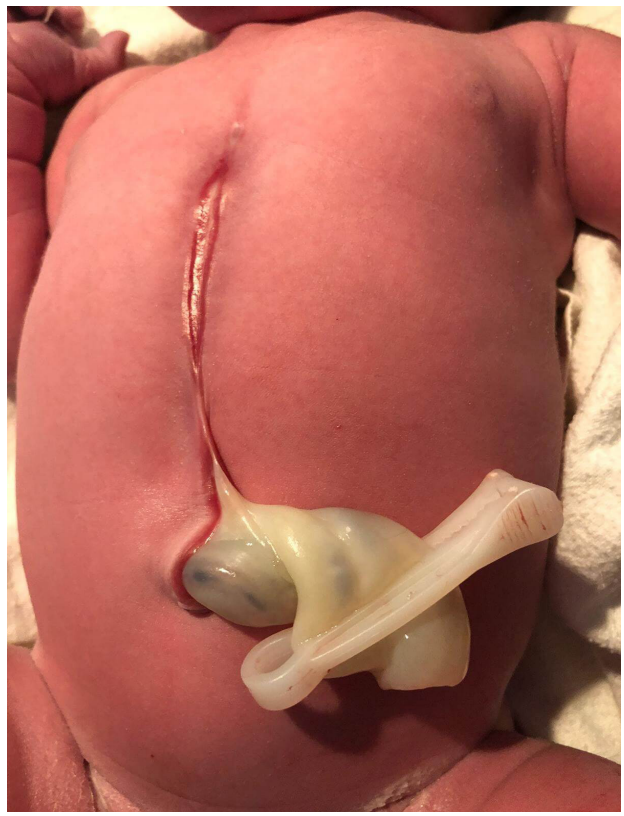
Figure 1. Incomplete superior sternal cleft with visible skin defect from the xiphoid process to the umbilical cord.



Sternal clefts can be divided into 3 categories: superior, inferior and complete.[6] This patient presented a medial cleft from the sternal body to the xiphoid process and a relevant distance between the sternal bars was seen only in the manubrium. Therefore, we consider this an incomplete superior sternal cleft, which is the most common type. This is not usually associated with cardiac defects and is more often related to supraumbilical raphae (present in our patient), cervicofacial hemangiomas and PHACE syndrome.[3,7] PHACE syndrome is a rare neurocutaneous disorder of unknown etiology and our patient meets the criteria for possible PHACES: the hemangioma

Figure 2. Thoracic computed tomography revealing the bifidity of the manubrium (arrow).

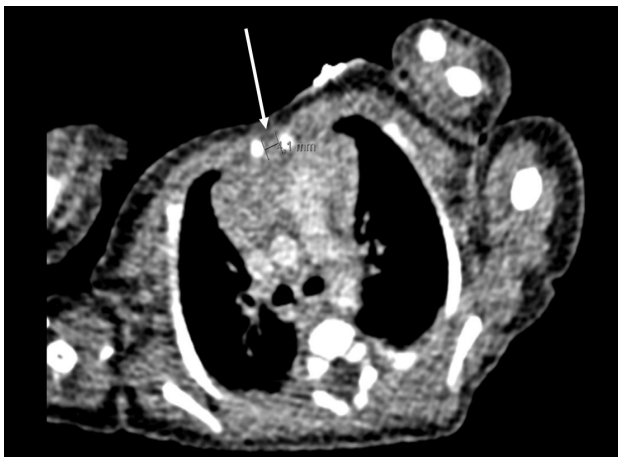
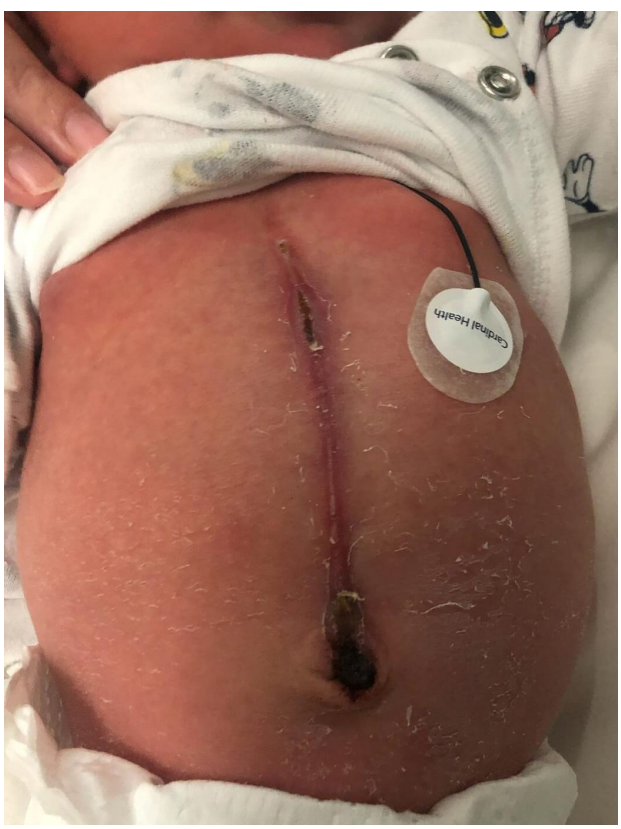


Figure 3. Partial epithelization of the skin defect by 10th day of life.



of the trunk and the midline sternal and abdominal defect.[4] Along with the echocardiogram, which was normal in this case, a magnetic resonance angiography of the head, neck, and aortic arch is also recommended in order to screen for the common cerebral vascular anomalies. Our patient is also waiting for an ophthalmologic examination, which is important for the assessment of possible congenital developmental anomalies, such as glaucoma or cataracts.[5] Although she does not fully meet the diagnostic criteria for definite PHACES syndrome, a regular follow-up is extremely important for the timely assessment of other possible associated conditions, such as hearing deficits, endocrine disorders, and dental anomalies.[8] Corrective surgery of the sternal cleft is considered the best treatment option as it will increase respiratory function, protect mediastinal structures in case of trauma and improve physical appearance of the patient.[6] The safest timing for the surgery is the neonatal period as the chest wall compliance will decrease the risk for cardiovascular injury.[9] Given the sustained clinical stability of our patient by three months old, without any signs of respiratory distress, she has not been submitted to any corrective surgery and will maintain close follow-up consultations. She has not presented with any other complications associated with PHACES syndrome.

Compliance with Ethical Standards

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Conflict of Interest None

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