A Pulsating Chest
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After watching the video clip (Video), further evaluation for this infant should include 1 or more of the following:
A. Echocardiography
B. Ophthalmologic examination
C. Renal ultrasonography
D. Skeletal survey
E. Upper gastrointestinal series

CRITIQUE
Case
The female infant in this video clip was born at 39 weeks’ gestation to a 40-year-old gravida 4 para 2 woman with normal prenatal findings. The infant was delivered via cesarean following a fetal heart rate tracing with variable decelerations. The infant required continuous positive airway pressure for 3 minutes. Her Apgar scores were 8 and 9 at 1 and 5 minutes of age, respectively. Her birth parameters included a weight of 3,700 g (84th percentile), length 53.0 cm (98th percentile), and occipitofrontal circumference 35.5 cm (91st percentile).

Her physical examination findings were notable for a sternal defect with pulsations. Because of this finding, she was transferred to the NICU, where she initially required supplemental oxygen via high-humidity nasal cannula, but was quickly weaned to room air within a few hours. The video clip was obtained shortly after the infant was admitted to the NICU. The clip depicts a newborn with a U-shaped defect in the superior aspect of the sternum with a membranous defect over the xiphoid. The sternum collapses posteriorly during inspiration. Pulsations can be seen under the membranous defect between respiratory efforts. The skin over the sternal defect is normal. The infant did not have any anterior abdominal wall defects and there were no other midline defects.

AUTHOR DISCLOSURE Drs Mammel and Vachharajani have disclosed no financial relationships relevant to this article. This commentary does not contain a discussion of an unapproved/investigative use of a commercial product/device.
The video findings are consistent with a partial superior type sternal cleft (SC). This is a type of chest wall malformation caused by a defect in the sternal fusion process. An SC is a rare congenital chest wall malformation with a reported incidence of less than 1% of all chest wall malformations. More common chest wall malformations include apectus excavatum (caved-in) and pectus carinatum (protrusion).

Sternal development begins during the 6th week of gestation with the formation of a pair of parallel mesenchymal bars. Between the 6th and 10th weeks of gestation, cells migrate from these 2 lateral bars to fuse and form a series of sternal bars in the midline. This process occurs in a craniocaudal direction. The manubrium arises from a central pre-sternal mass and bilateral supra-sternal masses. The pre-sternal mass fuses with the cranial aspect of the sternal bars; the supra-sternal masses develop into the sternocleidomastoid joints. An SC results from a defect at some point in the fusion process. Inferior SC is thought to result from a failure of the fusion of sternal bars, while a superior SC may result from failure of manubrium formation.

The physical examination of a newborn with an SC often shows a paradoxical midline thoracic bulging. Depending on the extent of the defect, an SC can be small and be undetected, or can be complete and associated with ectopia cordis. Infants with a partial SC are typically asymptomatic at birth, though affected infants are at increased risk of injury to the mediastinal structures, and can develop respiratory symptoms if the SC is not surgically corrected.

The phenotypic spectrum of SCs is broad; an SC can be partial or complete, superior or inferior, and can be isolated or associated with other defects. One review found that an SC was associated with other defects in 73% of cases (n = 63/86).

Initial evaluation of an infant with an SC should include a detailed physical examination to assess for hemangiomas, cleft lip or palate, skin tags, supraumbilical raphe, and an omphalocoele. If hemangiomas are found, the infant may require a laryngotraceobronchoscopy to rule out subglottic hemangiomas.

A superior SC can be a component of PHACES syndrome. PHACES stands for Posterior fossa malformations, Hemangiomas, Arterial anomalies, Cardiac defects, Eye abnormalities, and SC and supraumbilical raphe.

An ophthalmologic examination and posterior fossa imaging may be indicated in a patient with an SC, particularly if hemangiomas are identified. A cardiologic examination to assess for structural cardiac defects, as well as an echocardiography should be performed.

An inferior SC can be seen in association with pentalogy of Cantrell. The 5 anomalies found in patients with pentalogy of Cantrell consist of (a) anterior diaphragmatic defect, (2) midline supraumbilical abdominal wall defect, 3) defect in the diaphragmatic pericardium, 4) intracardiac abnormalities, and 5) defect of the lower sternum. A diagnosis of complete pentalogy of Cantrell is rare and associated with a high mortality.

The infant in this video was diagnosed as having an SC immediately after birth. Physical examination did not reveal any hemangiomas. Brain magnetic resonance imaging showed a normal posterior fossa. Echocardiography revealed a small patent ductus arteriosus, which had closed on repeat imaging 2 days later. The neonate also displayed dilation on ophthalmologic examination, which was found to be normal. The infant’s chromosomal microarray analysis was normal. Thus, the infant had an isolated partial superior SC. At 3 days of age, she underwent primary closure without hemodynamic or respiratory compromise. Postoperatively, she was observed in the cardiac intensive care unit for 44 hours and then transferred back to the NICU before discharge.

CORRECT RESPONSES

A and B. Based on the clinical findings of a superior partial SC, the infant’s evaluation should include echocardiography to rule out structural cardiac defects, as well as an ophthalmologic examination to assess for eye abnormalities associated with PHACES syndrome. The infant did not have any hemangiomas on physical examination, which would raise the index of suspicion for PHACES; however, given the high rate of defects associated with an SC, it is reasonable to perform a complete evaluation.

References
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