PHACES syndrome is a spectrum of anomalies, P, posterior fossa anomalies as Dandy–Walker malformation; H, hemangioma; A, arterial lesions of the head and neck (the most commonly detected include dysplasia, aberrant origin or course, hypoplasia, and absence or agenesis); C, cardiac abnormalities as aortic coarctation; E, abnormalities of the eye and S, sternal defect, that may be present in up to 2% of children with facial hemangiomas and 20% of children with segmental facial hemangiomas. The constellation of PHACES syndrome symptoms may vary significantly between different patients. Major and minor criteria for PHACES syndrome have been recently described in order to improve their classification and management. We report the case of a newborn with PHACES syndrome, who had additional congenital defects including ectopia cordis as the most severe form of midline defect. Although the list and variety of published cardiac malformations in PHACES syndrome are extensive, ectopia cordis has not been previously reported.

1. Introduction

Complete sternal cleft with ectopia cordis is a rare visually dramatic congenital anomaly that results from the failed ventral migration and fusion of the two lateral mesodermal sternal bands between the sixth and ninth weeks of gestation. The cause of sternal cleft is unknown and most of them occur sporadically. Sternal cleft can be classified as superior, inferior, or complete. Superior sternal cleft is the most common variant associated with the PHACES syndrome. Complete forms with ectopia cordis have not been previously reported suggesting a further expansion of the spectrum of the syndrome.

2. Case report

Our patient was a female infant born during the 39th weeks’ gestation without an antenatal diagnosis of ectopia cordis or Cantrell’s syndrome. The postnatal examination confirmed the diagnosis (Fig. 1). The anterior thoracic defect extended from the neck to the lower border of a small sternum at the level of the fourth rib only covered by thin skin that allowed the cardiac impulse to be seen. The sternum was entirely deficient and the heart was extra-thoracic oriented. There was a normal arrangement of the great vessels and an intact diaphragm. The pericardium was reconstructed and the heart relocated within the thoracic cavity without hemodynamic compromise. The postoperative period was uneventful.

At 20 days of age, a sudden onset of a large red macule on the face with a segmental distribution on the mandibular area was noticeable. The lesion was mainly on the left side of the face, but the right side was also involved (Fig. 1). Supraumbilical raphe and mild retrognathia were also observed. The macule rapidly enlarged to form a vascular plaque. Brain and thorax imaging [computed tomography (CT) angiogram of the brain, neck and thorax] were performed showing an aberrant communication between left common carotid and subclavian arteries without persistence of trigeminal artery or posterior fossa malformations (Fig. 1).

Follow-up at six-month of age showed a good cosmetic result and a well-tolerated repair.

3. Discussion

Thoracic ectopia cordis is extremely rare with a reported incidence of 5.5–7.9/million live births. In partial thoracic ectopia cordis the heart can often be seen to pulsate through the skin. In complete thoracic ectopia cordis the naked heart is displaced outside the thoracic cavity without pericardial coverage. Many cases of this type have an associated intra-cardiac defect including ventricular septal defect, atrial septal defect, tetralogy of Fallot, left ventricular diverticulum and pulmonary hypoplasia [1].

Complete thoracic ectopia cordis left untreated is universally fatal. The aims of surgical treatment are: to provide soft tissue cover of the heart, to reduce the heart into the
thoracic cavity; palliation or repair of any intra-cardiac defect and reconstruction of the chest wall.

Reduction of the heart into a small thoracic cavity in the neonatal period often produces compression, kinking of the great vessels and a low cardiac output. However, it is worthwhile making an attempt at placing the heart even partially within the thoracic cavity at the first stage. This would make subsequent procedures easier and also avoid the obvious physical deformity. The heart can be returned to the left or right chest depending on which way the apex points.

A subgroup of patients with infantile hemangiomas have associated structural anomalies of the brain, cerebral vasculature, eyes, sternum, and/or aorta in the neurocutaneous disorder known as PHACES syndrome. Females are affected more often than males by a ratio of 9:1. This disparity is higher than infants with large cervicofacial segmental hemangiomas associated with PHACES syndrome. The diagnosis has been broadly inclusive by using a case definition of a facial hemangioma plus one extracutaneous features, leading to numerous reports of potential associated disease features, many of uncertain significance [2–6]. Cleft sternum, and other midline defects belong to the criteria of PHACES. Therefore, the occurrence of ectopia cordis and supraumbilical raphe associated to segmental hemangioma of the face, should be considered part of the syndrome.

A consensus statement was thus developed to establish diagnostic criteria for PHACES syndrome. A multidisciplinary group of specialists with expertise in PHACES syndrome drafted initial diagnostic criteria on the basis of the review published, peer-reviewed medical literature and clinical experience during the PHACES Syndrome Research Conference held in Texas in November 2008 [7]. Major and minor criteria were determined for the following organ systems: cerebrovascular, structural brain, cardiovascular, ocular, and ventral/midline.

Definite PHACES requires the presence of a characteristic segmental hemangioma or hemangioma > 5 cm on the face or scalp plus one major criterion or two minor criteria. Possible PHACES requires the presence of a hemangioma > 5 cm on the face or scalp plus one minor criterion.

More than 20 different cardiovascular anomalies have been previously reported in children with PHACES syndrome.

Up to our knowledge, this is the first report of a newborn with ectopia cordis as a cardiac anomaly in the context of PHACES syndrome.

References

