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Case Report

PHACES syndrome with Ectopia cordis

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ABSTRACT

PHACES syndrome is an acronym for, P, posterior fossa anomalies as Dandy- Walker malformation; H, hemangioma (capillary segmental faces); A, arterial lesions of the head and neck (the most common ones include aberrant origin or course, hypoplasia, dysplasia and agenesis); C, cardiac abnormalities as coarctation of aorta; E, abnormalities of the eye and S, sternal defect. The constellation of findings of PHACES syndrome may vary significantly in different patients. Complete sternal cleft with ectopia cordis is an extremely rare congenital anomaly failed ventral migration and fusion of the two-lateral mesodermal sternal bands between the sixth and ninth weeks of gestation. We are reporting a rare case of PHACES syndrome, with partial ectopia cordis as a midline defect, who is in our follow up since birth. Survival of the child without surgical intervention with epithelialization over the defect and Leucomatous corneal opacity are interesting findings.

Keywords: Ectopia cordis, Hemangioma, Leucomatous corneal opacity, PHACES syndrome

INTRODUCTION

PHACES syndrome is coexistence of anomalies, P, posterior fossa anomalies as Dandy-Walker malformation; H, hemangioma (capillary segmental faces); A, arterial lesions of the head and neck (the most common ones include aberrant origin or course, hypoplasia, dysplasia and agenesis); C, cardiac abnormalities as coarctation of aorta ; 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.[1] The constellation of findings of PHACES syndrome may vary significantly in different patients. Complete sternal cleft with ectopia cordis is a rare congenital anomaly due to failed ventral migration and fusion of the two-lateral mesodermal sternal bands between the sixth and ninth weeks of gestation.[2] We are reporting a rare case of PHACES syndrome, with partial ectopia cordis as a midline defect, who is in our follow up since birth. Survival of the child without surgical intervention with epithelialization over the defect and Leucomatous corneal opacity are interesting findings.

CASE REPORT

This female child was born full term to Muslim parents out of consanguineous marriage with ectopia cordis and absent sternum and otherwise unremarkable birth history (Figure1). 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 membrane that allowed the cardiac impulse to be seen. The sternum was entirely deficient and the heart visible through the defect. 2D Echo was suggestive of ectopia cordis with severe coarctation of arch of aorta with left ventricular hypertrophy. After stabilisation, child was referred to CTVS and paediatric surgery departments to provide soft tissue cover to the heart and reconstruction of the anterior chest wall so as to avoid the obvious physical deformity

and advised application of antiseptic ointment due to refusal of parents for surgical intervention.

Figure 1: Ectopia cordis at birth.

Child was brought at 18 months of age with complaints of fever, cough, and difficulty in breathing and decreased oral acceptance and diagnosed as bronchopneumonia with respiratory distress and failure to thrive. Child has history of bottle feeding with diluted top milk.

Figure 2: Epithelialisation over sternal defect.

On examination her weight was 8 kg, pallor was present with tachycardia (HR: 142/min) and tachypnoea (RR: 62/min) with SPO2 88% at room air and 98% with oxygen. Small haemangioma on the face with a segmental distribution over the mandibular area was noticeable on left side of face and a large hemangioma of 6 cm was noted on chest. Sternum was absent, and heart was covered with thin skin and soft tissues with cardiac impulse visible. Cataract and Leucomatous corneal opacity were noted in left eye on ophthalmic reference. Bilateral equal air entry with bilateral crepts and wheeze was present (Figure 2).

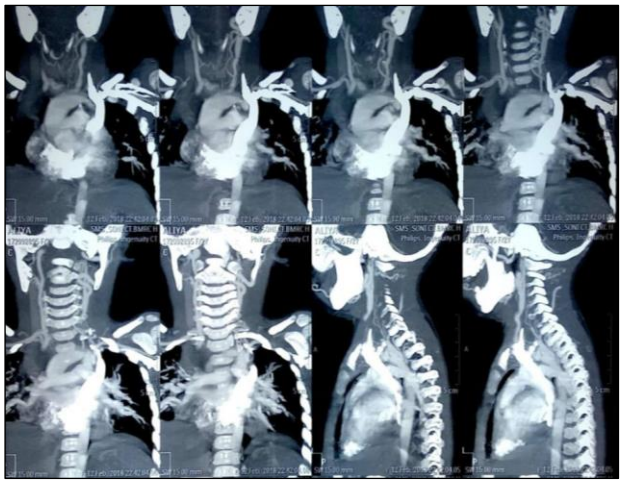

Figure 3: CT Carotid angiography.

CBC was suggestive of microcytic hypochromic anemia. H1N1 was negative. Pulmonary angiography and CT Cerebral angiography were normal. CT carotid angiography depicted that there is left common carotid artery and internal carotid artery is severely narrowed in caliber with reformation of left terminal ICA/ACA-MCA through collateral circulation (Figure 3).

DISCUSSION

Thoracic ectopia cordis is very rare with a reported incidence of 5.5-7.9/million live births. It is currently believed that the PHACES is a consequence of some unknown insult at an early stage of fetal development. Sternal cleft can be classified as superior, inferior, or complete. Superior sternal cleft is the most common variant associated with the PHACES syndrome. 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 complete ectopia cordis have an associated intra-cardiac defect including ventricular septal defect, atrial septal defect, tetralogy of Fallot, left ventricular diverticulum and pulmonary hypoplasia.[1]

Hemangiomas are the most common, benign tumors occurring in 4-5% of children.[2] Hemangiomas of the face and neck are associated with structural defects of the central nervous system, including cerebral vascularity and defects of the heart, aorta and eyes.[3] As in this case; hemangiomas are often absent at birth. The coexistence of extensive facial and neck hemangiomas with intracranial disorders of the brain and vascular system structure was first described in 1978 by Pascual-Castroviejo.[4] In 1996, Frieden and colleagues proposed to name the complex of accompanying defects, as acronym PHACE.[5] Brain structural disorders occur in about 50% of patients with confirmed PHACES syndrome.[6,8] A Research conference held in Texas in November 2008, determined major and minor criteria for the following

organ systems: cerebrovascular, structural brain, cardiovascular, ocular, and ventral/midline.[7] Definite PHACES requires the presence of a characteristic segmental hemangioma or hemangioma of 5 cm on the face or scalp plus one major criterion or two minor criteria. Currently, diagnosis of the PHACES syndrome requires the co-existence of a segmental haemangioma or haemangioma with a diameter of more than 5 cm and one or more of the lesser defects characteristic of the syndrome. In the present case CT angiography brain was normal, presence of Hemangioma and sternal defect and eye abnormalities in our case with coarctation of aorta is in consonance with the diagnosis of PHACES.

Disorders of the cardiovascular system occur in as many as 67% of children.[8] The most often described is coarctation of the aorta.[9] Relatively often, there is a right-sided aortic arch, double or complete disruption of the aortic arch with subsequent significant aneurysm widening of adjacent sections, as well as abnormal course and point of withdrawal of subclavian arteries.[9] Coarctation of the aorta often occurs in the general population and accounts for 5-8% of congenital heart defects. The defects of the interventricular septum are slightly more frequent in the PHACES syndrome and therefore they were considered as a smaller diagnostic criterion. Our case has coarctation of Aorta.

Impaired vision results from abnormal visual stimulation and may be a consequence of squinting, hyperopia (anisometropia), high bilateral refractive error (isometropia) and deprivation of visual stimuli (eyelid drooping, cataract).[10] Ophthalmic reference in our patient revealed corneal opacity (leucomatous) and cataract in left eye.

Complete thoracic ectopia cordis left untreated is universally fatal. Only a dozen of examples of total sternal cleft have been published in the world literature.[11]

CONCLUSION

We are reporting a case of Partial ectopia cordis who was in our follow up since birth. Hemangioma not present at birth appeared at 1 month of age and is visible now also. Survival of the child without surgical intervention with epithelialization over the defect and Leucomatous corneal opacity are unique findings.

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