PHACE(S) syndrome in a 8 month old girl

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Abstract

TA case of haemangioma of infancy associated with coarctation of the aorta occurring in a 8 month old baby girl from Mathugama is reported. The haemangioma was multiple plaque type distributed on the left side of upper face and head. Absent femoral pulses and a systolic murmur on the left sternal edge suggested coarctation of the aorta. This was confirmed by echo cardiography and CT scanning.

Introduction

Haemangiomas of infancy are rarely associated with malformations elsewhere. The most well documented entity is the PHACE(S) syndrome. The acronym stands for P - posterior fossa malformations, H - haemangioma, A - arterial abnormalities, and C - coarctation of aorta, E - eye abnormalities, S - sternal defects. To date this neurocutaneous syndrome has occurred mainly in girls.

Case report

And eight-month-old baby girl presented in August 2004 to the dermatology unit at LRH for evaluation of a haemangioma of the face. The lesions were first observed at the age of 4 weeks. She is the third child of a non-consanguinous marriage. At birth the child had been fully assessed at the local general hospital. Her antenatal and post natal periods were unremarkable and was otherwise healthy, achieving appropriate milestones for her age.

Examination revealed an active child whose weight and head circumference were appropriate for the age. She had no dysmorphic features. She had a large haemangioma, made up of multiple plaques involving the upper eye lid, temporal region, helix of the ear and the occipital region on the left side. There was partial closure of the left eye, but no visible ocular defects (figure 1).

Examination of the cardiovascular system revealed a right radial pulse of good volume with a regular 80 beats per minute rate, while the left radial pulse was weak and thready. Femoral pulses could not be felt. The blood pressure on the right arm was 110/60 mm Hg, while the pressure on the left arm was 80/60 mm of Hg. An ejection systolic murmur of grade 2 severity was heard at the aortic area. There was no evidence of cardiac failure. Other systems were clinically normal.

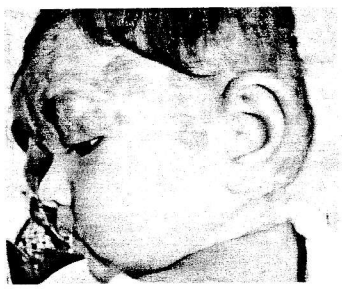

Figure 1.

In view of the known association and absent femoral pulses with a blood pressure difference on the upper limbs, coarctation of the aorta was strongly suspected. An echo cardiogram by consultant paediatric cardiologist confirmed the presence of coarctation with a pressure gradient of 53 mm Hg with mild left ventricular hypertrophy and no evidence of septal defects. CT angiogram of the chest and lower neck showed a markedly tortuous arch of the aorta which extended cranially with a narrowed segment after origin of the left subclavian artery (figure 1).

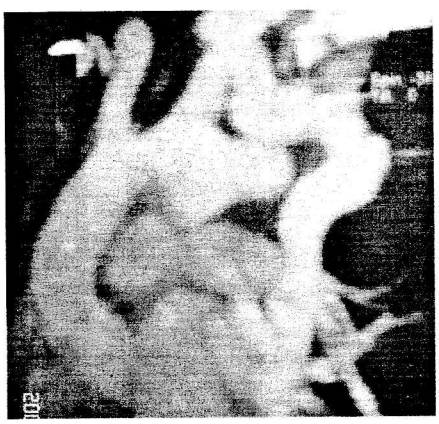

Figure 2.

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Ultrasonography and contrast enhanced CT brain did not reveal any abnormalities in the brain including the posterior fossa. Ophthalmological assessment did not reveal any abnormality.

Discussion

Haemangioma are the most common benign tumours of infancy. These lesions may be present at birth but more often arise during the first few weeks of life like in this patient. Haemangioma are more commonly found in girls than in boys. Haemangiomas show considerable variation in appearance, anatomical location, number, depth and stage of evolution.

Segmental haemangiomas are relatively less common and generally plaque like lesions arranged in linear or geographic manner, present over specific cutaneous territories. These segmental haemangiomas are much more likely to occur in Hispanic infants and infants of greater birth weight or gestational age. Further more segmental haemangiomas are more likely to be associated with complications such as ulceration developmental anomalies and visceral haemangiomas; require more intensive and prolonged therapy, and have a poor overall outcome.

Structural abnormalities are rarely associated with haemangiomas. Only a few such syndromes have true haemangiomas as a cardinal feature. Among the well documented entities PHACE(S) syndrome is unique. To date majority of the reported cases are females (88%). Often affected girls show one or two of the associated features rather than all components. Any girl with a large upper facial segmental haemangioma should be examined thoroughly for signs and symptoms of PHACE(S) syndrome, including a careful cardiac evaluation, cardiac ultrasound and measurement of blood pressure in all four limbs and careful periodic ophthalmological and neurological assessment.

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References

1. Frieden IS, Reese V, Cohen D. PHACE syndrome. The association of posterior fossa brain malformations, heamangiomas, arterial anomalies, coarctation of aorta and cardiac defects, and eye abnormalities. *Arch Dermatol* 1996; **132**: 317-21.
2. Metry DW, Dowd CF, Barkovich AJ, et al. The many faces of PHACE syndrome. *J.Paediatr* 2001; **139**: 117- 23.