CASE REPORT

PHACE syndrome with lip haemangioma, microphthalmos and persistent fetal vasculature

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SUMMARY

An 11-month-old baby girl presented with white reflex in her left eye. On examination, there was a 6.5×5 mm² haemangioma present over her face involving on her lower lip. Systemic examinations were within normal limits. The left eye was small, with an axial length of 16.08 mm and had a cataract. Ultrasonography of the left eye was suggestive of the presence of a vascular stalk, persistent hyperplasia of a primary vitreous, or persistent fetal vasculature with vitreous haemorrhage. On MRI, the left eye was small with vitreous haemorrhage. Left eye lens aspiration was performed and the bleeding vascular stalk behind the lens was cauterised with diathermy. The right eye was normal. The patient was diagnosed as having PHACE syndrome (Posterior fossa malformations, Hemangiomas, Arterial anomalies, Coarctation of the aorta and other cardiac defects, and Eye abnormalities syndrome). On follow-up, she was able to follow light with her left eye.

BACKGROUND

Posterior fossa malformations, Haemangiomas, Arterial anomalies, Coarctation of the aorta and other cardiac defects and Eye abnormalities (PHACE) syndrome represents the association of large infantile haemangiomas of the head and neck with brain, cerebrovascular, cardiac, ocular and ventral or midline defects. PHACE syndrome is definitely diagnosed with presence of an infantile segmental 5 cm² haemangioma of the head (scalp and face) with one major or two minor criteria.¹ In our patient, one major criterion, persistent hyperplasia of a primary vitreous (PHPV) and two minor criteria, microphthalmos and cataract, were present. There were neither central nervous system nor other system anomalies present in this patient. Although facial haemangioma in PHACE syndrome has previously been reported,² a large segmental haemangioma affecting only the lip and ocular manifestation in PHACE syndrome has not been reported yet, to our knowledge. We report a unique case of a large lip haemangioma with ocular malformations in PHACE syndrome without any other systemic malformation, where we operated on the patient for cataract and prescribed the proper power of glasses.

CASE PRESENTATION

An 11-month-old baby girl presented with white reflex in her left eye. On examination, there was a 6.5×5 mm² haemangioma present over her face involving on her lower lip (figure 1). Systemic examination findings were within normal limits.

The baby weighed 8 kg, appropriate for her age. There was no developmental delay. She was following light with her right eye but resisted her right eye occlusion for left eye examination. On ocular examination, the left eye was small, with an axial length of 16.08 mm and had a cataract. The right eye was normal. The baby was the parents' first child and there was no family history of a similar presentation. There was no history of drugs taken by the mother during the gestational period and the birth history was unremarkable.

INVESTIGATIONS

Ultrasonography (USG) was performed to rule out any posterior segment pathology. On USG, there were two moderate to high spikes, one in the vitreous cavity and the other from an elevated lesion on the optic disc, with some mild amplitude spikes in the vitreous cavity, suggestive of a vascular stalk of PHPV or persistent fetal vasculature (PFV) with associated vitreous haemorrhage (figure 2). MRI of the head, neck, chest and orbit were performed to

Figure 1 An 11-month-old baby girl with a 6.5×5 mm² haemangioma on her lower lip and a small left eyeball (microphthalmos).

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Figure 2 Ultrasonography showing two moderate to high spikes, one in the vitreous cavity and the other in an elevated lesion of the optic disc with some mild amplitude spikes, suggestive of a vascular stalk of persistent hyperplasia of primary vitreous with vitreous haemorrhages.

rule out any space occupying lesion or malformation. On MRI, the left eye appeared small with signs of a cataract and vitreous haemorrhage (figure 3).

DIFFERENTIAL DIAGNOSIS

The patient was diagnosed as having PHACE syndrome with presence of facial haemangioma larger than 5 mm and one major criterion of ocular involvement, PHPV and two minor criteria, microphthalmos and cataract.

Figure 3 MRI scan showing a small left eyeball with a more radio-opaque lesion in the anterior segment of the eye and a diffuse mild hyperechoic lesion in the vitreous cavity suggestive of cataract with haemorrhages in the vitreous cavity.

TREATMENT

Left eye lens aspiration was planned to clear the media and after the lens aspiration was performed by bimanual irrigation-aspiration system, a membrane became visible behind the lens, with a bleeding vascular stalk, showing features of PHPV, which was cauterised with monopolar diathermy. The vascular stalk arose from the optic disc up to the membrane behind the lens. After cauterisation of one end of the vascular stalk, it regressed back to the vitreous cavity. As the eyeball was small, the intraocular lens was not implanted, leaving the patient left aphakic. Refraction was carried out and a +19.00 diopter left eye aphakic glass was given postoperatively with advice of occlusion therapy 1:1 in the right and left eye, respectively, to prevent amblyopia.

Systemic therapy with propranolol 1 mg/kg body weight/day was started for the lip haemangioma after consulting the paediatrician.

OUTCOME AND FOLLOW-UP

On next postoperative day, the media was clear. Fundus glow was present. On seventh postoperative day, the baby was following light and was not resisting the right eye occlusion. The media was clear with good fundus glow. The lip haemangioma decreased after 1 month of propranolol therapy and the baby continued the propranolol.

DISCUSSION

Infantile haemangioma is the most common benign tumour of infancy, with an estimated incidence of 4% to 5%.³ A subgroup of patients with infantile haemangiomas exhibit associated structural anomalies of the brain, cerebral vasculature, eyes, aorta and chest wall in the neurocutaneous disorder, PHACE syndrome (OMIM 606519). The PHACE acronym was coined by Frieden *et al*,⁴ in 1996. Ninety-eight per cent of published cases of PHACE syndrome report a haemangioma located on the head. The face is the next location, with segmental distribution

Patient's parent's perspective

My baby girl was operated on in her left eye for cataract but an intraocular lens was not placed due to the small size of the eye. Now she is at least following light. We are happy with the intervention and would like to follow-up regularly.

Learning points

- ▶ Ocular examination should be performed in addition to other system evaluation in patients with facial haemangioma and microphthalmos.
- ▶ Ultrasonography should be performed if there is a cataract, to rule out any posterior segment pathology such as persistent hyperplasia of a primary vitreous, coloboma, peripapillary staphyloma, etc.
- ▶ MRI of the head, neck and chest should be performed to rule out any space-occupying lesion or malformation. Intracranial and cervical vessel MR angiography is advised to rule out vascular malformation.

of the haemangioma. Cannady *et al*⁵ reported a case of a patient who presented with respiratory distress at birth, secondary to a large nasal glioma. That patient was subsequently found to have a ventricular septal defect a facial haemangioma and a malformation of the eye and optic nerve head. Lasky *et al*⁶ reported a case of PHACE syndrome in association with PFV and coloboma-like iris defect. Ophthalmological findings have been noted in approximately one-third of patients reported with PHACE syndrome. Reported ophthalmological abnormalities include choroidal haemangiomas, cryptophthalmus, exophthalmus, colobomas, posterior embryotoxon, optic atrophy, microphthalmos, strabismus, congenital glaucoma and optic nerve hypoplasia.⁷

Our case was a unique presentation, with a large haemangioma of the lower lip with no other involvement of the head and face and associated ocular malformation only.

Our patient presented late. Otherwise, the cataract might have been operated on early to prevent amblyopia in the affected eye.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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