A Congenital Glaucoma Associated with Phakomatosis Pigmentovascularis in Infant Case Report

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ABSTRACT

Phakomatosis Pigmentovascularis (PPV) is the rare condition which has been classified in the same spectrum with Sturge-Weber syndrome, Klippel-Trenaunay-Weber syndrome and oculodermal melanocytosis.1 PPV is the combination of widespread vascular lesions and extensive pigmentary lesions. We report a 2-month-old-infant with PPV type IIa associated with congenital glaucoma. She showed extensive Port-wine stain, extensive Mongolian spots and Café au lait spots along with soft tissue hypertrophy on her right face. She had buphthalmos on her right eye and the very high intraocular pressure, so she was diagnosed as congenital glaucoma.

Keywords: Glaucoma; phacomatosis pigmentovascularis; nevus flammeus; Mongolian spots; ocular melanocytosis (Siriraj Med J 2017;69: 217-219)

INTRODUCTION

Neural crest cell disorder can result in a variety of syndromes including PPV. PPV which were first described by Ota et al.[1] This very rare cutaneous condition has happened sporadically, most cases were Asian and were classified into 4 subgroups.[3] Ocular abnormalities associated with PPV have been reported[4-5] including glaucoma, melanosis oculi, prominent vessel in sclera, iris mammillations, choroidal hemangioma, choroidal melanocytosis/melanoma, and optic nerve melanocytoma.

CASE REPORT

A 2-month-old-infant presented with reddish discoloration on right side of her face. Her mother was P_2A_0 with no underlying disease. Her brother was healthy with no birthmark. Her birth weight was 2,870 grams with normal labour, and she stayed in hospital for 3 days before she returned home with no problem. She had normal development, with no history of convulsion. The pediatrician in her nearby hospital detected her extensive birthmark and buphthalmos on her right eye and referred her to Siriraj Hospital. At Siriraj, the ophthalmic examination revealed visual acuity fix and follow both eyes. Intraocular pressures were 33 and 18 mmHg in her right and left eye respectively. Corneal diameters were 12 mm (vertical), 12 mm (horizontal) on her right eye while 10.5 mm (vertical). 11 mm (horizontal) on her left eye. The right showed buphthalmos with slate-gray scleral discoloration with slightly dilated episcleral vessel on the nasal side of the eye. Gonioscopy revealed open angle, lightly pigmented angle structure with no blood in schlemm's canal in both eyes. No iris mammillations were found. Lens were clear in both eyes. Fundi showed normal color with no choroidal hemangioma detected. There were sharp, pink optic nerve heads in both eyes with enlarged cupping in her right optic disc. The vertical cup to disc ratios were 0.4, 0.2 in her right and left eye respectively. A congenital glaucoma was diagnosed on her right eye. The skin examination showed erythematous patch on the right side of the face, scalp, back, chest, arms, right leg and greenish-blue patches on trunk, arms and legs. She had soft tissue hypertrophy on her right face and

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Café au lait spots on her right buttock. This patient was diagnosed as PPV type IIa or Phacomatosis cesioflammea which is defined by nevus flammeus and Mongolian spots with no systemic findings. For glaucoma, the patient received topical medications which were latanoprost eye drop once before bedtime and brinzolamide eye drop twice a day on her right eye. The intraocular pressure was decreased from 33 to 18 mmHg in her right eye at 3-month-visit. After 3 month appointment she lost her follow-up and came back again 6 months later with uncontrolled intraocular pressure and more enlarged vertical cupping, from 0.4 to 0.8 in her affected eye.

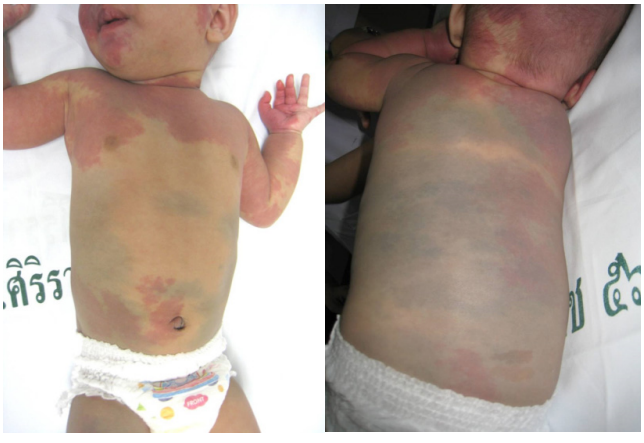

Fig 1&2. The vascular skin lesion was shown in this figure as an extensive nevus flammeus on her right face, trunk, arm, scalp and back. The pigmentary skin lesion was shown as an extensive Mongolian spots on her trunk and legs. Café au lait spots on her right buttock.

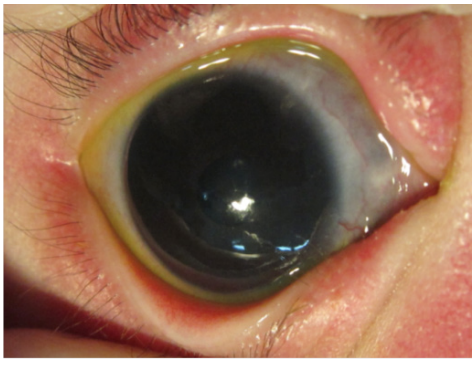

Fig 3. Buphthalmic eye with ocular melanocytosis, dilated episcleral vein and port wine satined involve upper and lower eyelid.

DISCUSSION

PPV is the cutaneous disorder which has been reported associated with vascular, neurologic and ocular abnormalities.[4] Congenital glaucoma can be found in oculodermal melanocytosis, Sturge-Weber (SW) syndrome, Klippel-Trenaunay-Weber (KTW) syndrome and PPV. There are 3 theories which describe the pathophysiology of glaucoma in PPV. First is the developmental anomaly of the angle[1] because trabecular meshwork is derived from neural crest cell and there is evidence of multiple anomalous angle structures which have been reported in SW and KTW syndrome with congenital glaucoma. Second is an increase episceral venous pressure because there were several reports that 30-70% of SW patients also suffered from glaucoma.[6] It has been proposed that trabecular dysgenesis plays an important role in glaucoma which occurs in the first two years of life whereas raised episcleral venous pressure is the main mechanism when glaucoma occurs later in life.[7] The third is direct infiltration of angle by melanocytes.8 The largest literature which comprised of 9 cases of PPV patients who had glaucoma in Thailand stated that the strong predisposition for congenital glaucoma is extensive oculodermal melanocytosis and nevus flammeus which occur together involving the globe.[1]

In this patient, the examination revealed slate-gray scleral discoloration with slightly dilated episcleral vessel which was evidence of pigmentary and vascular lesion involving the globe as the strong predisposing factor for congenital glaucoma. The literature has said that surgical management is the mainstay of congenital glaucoma management, but sometimes we start from medication first. Although the topical medication is not an effective choice in treating congenital glaucoma, the parents had a tendency to choose this one first. She received topical Prostaglandins eye drops (Latanoprost) which showed favorable safety and efficacy profiles from RCT.[9] within contrast, Beta-adrenergic blockers may have a systemic side effect such as bradycardia and hypotension in young infants. The second line topical medication is carbonic anhydrase inhibitors with less systemic side effect and good efficacy. After she received these medications, she had partial response with decreased intraocular pressure, but it was not enough to control the disease progression. The parent was convinced to give the permission for angle surgery. Goniotomy, which has proved to have favorable outcome in the patient aged 1-24 months old, can be done more than 1 time with increased success rate.[10] The patient **underwent a Goniotomy**. She came for 1-week-after the operation follow-up visit with intraocular pressure of 29, and 14 mmHg on her right and left eye respectively. 2% Betoptic-S eye drop was added and second Goniotomy operation planned, but after that time she was lost to follow-up. As PPV is not the disease that affects one single organ, she was sent for pediatric and dermatology consultation at the first visit. The consultant pediatrician took a thorough medical and developmental history and it came out normal. The physical examination taken revealed the

skin lesion as described above with no abnormalities in musculoskeletal, nervous and other systems. PPV skin lesion has rare tendency to malignant transformation and requires no treatment. The cosmetic disfiguring from nevus flammeus is an important aspect to be concerned because it can have a great effect on self-esteem of the patient. The pulse dye laser has a favor outcome on this kind of vascular malformation.[11] In treating this multi-organ syndrome a multi-disciplinary team is needed.

This case showed a highlight that when a congenital glaucoma is found, one should look for an associated skin lesion. The syndrome needs life-long examination and special treatment from multi-disciplinary team.

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