CASE REPORTS
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A CASE OF PHAKOMATOSIS PIGMENTOVASCULARIS TYPE IIB WITH SEIZURES

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Abstract

Phakomatosis pigmentovascularis (PPV) is a genetic disorder characterized by association of capillary malformation with pigmentary nevi. We hereby report a rare presentation of PPV with Sturge-weber syndrome (SWS), Klippel-trenaunay syndrome (KTS) and Nevus of Ota (PPV typeIIb) in a 7 - year old female child.

Key words: Phakomatosis pigmentovascularis, Sturge weber syndrome, Klippel trenaunay syndrome, nevus of Ota, mongolian spots

Introduction

Phakomatosis pigmentovascularis (PPV) is a rare genetic disorder characterized by association of capillary malformation with pigmentary nevus. Five types of PPV are described with further subtype 'a' for cutaneous involvement only and subtype 'b' for cutaneous as well as systemic involvement.[1,2] Sturge-Weber syndrome (SWS) is a neurological disorder characterized by facial capillary malformation with ipsilateral ocular and brain anomalies.[3] Klippel-trenaunay syndrome (KTS) is defined as limb capillary venous malformation (CVM) associated with progressive overgrowth of the affected extremity and anomalies of venous system.[4] Nevus of Ota are bluish, patchy, dermal melanocytosis that affects the sclera and the skin around the eye.

Case Report

A 7-year old girl child came to outpatient department with complaints of asymmetry of left half of body and red patch on left side of face since birth along with history of seizures. She was full term vaginal delivery in hospital with birth weight of two kilograms. Her developmental milestones were normal. Her body weight was 15.7 kgs. and height was 108 cms. Examination revealed non-blanchable erythematous patch of port wine stain on left side of face in the distribution of all V1, V2 and V3 branches of trigeminal nerve (Fig.1). Multiple aberrant mongolian spots were present on trunk and back (Fig.2).

The patient had hypertrophy of left side of body with enlargement of left half of face, left lower limbs and left half of genitalia (Fig. 1,3,4,5). There was engorgement of veins on left lower abdomen crossing the mid-line. Higher mental functions including speech were normal. She had limping gait and motor examination was normal. The eye examination revealed bluish discoloration of sclera on both side consistent with Nevus of Ota. (Fig.5).

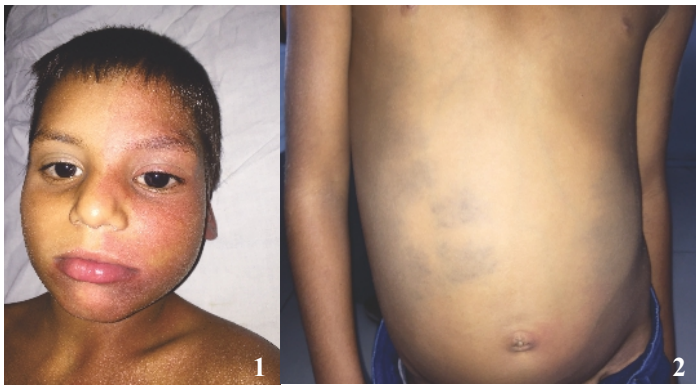


Figure 1 & 2: 1: Shows port wine stain and Hypertrophy of left half of face. **2:** Shows aberrant multiple mongolian spots.

Magnetic resonance imaging (MRI) of the brain showed cerebral hemiatrophy on left side with loss of white matter more significant in temporo-parieto-occipital region with mild peritrigonal FLAIR hyperintensity (Fig.6). Color doppler studies of lower limbs showed chronic thrombosis of left deep venous system with formation of superficial collaterals in left

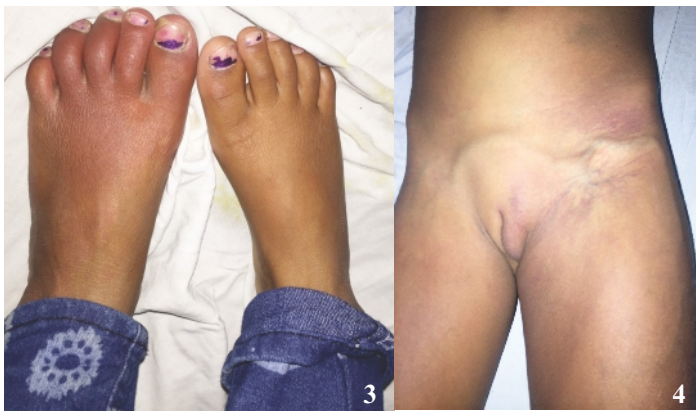
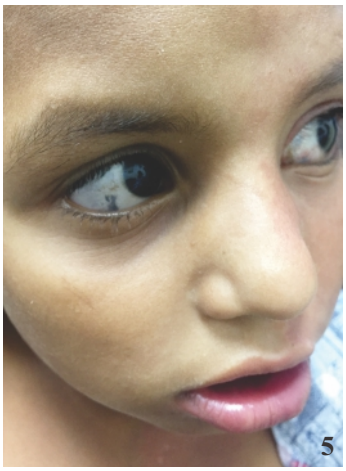


Figure 3 & 4: 3: Shows gross enlargement of left foot. **4:** Shows hypertrophy of left half of external genitalia & engorgement of veins.

inguinal region and upper part of left thigh. Right lower limb venous system was normal. Ultrasonography of abdomen was normal. Complete blood count, bleeding & clotting profile, liver function tests, renal function test were normal.



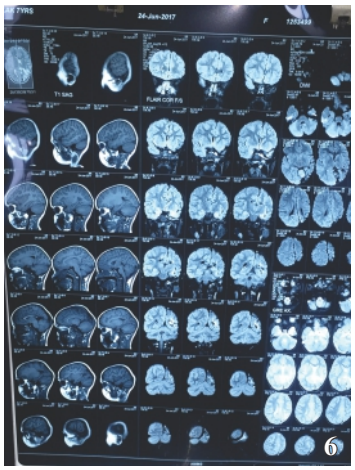


Figure 5 & 6: 5: Shows nevus of ota in both eyes. **6:** MRI Brain shows cerebral hemiatrophy on left side with loss of white matter more significantly in temporo-parieto-occipital region with mild peritrigonal FLAIR hyperintensity.

Discussion

Ota et al[1] in 1947 coined the term "Phakomatosis pigmentovascularis" and reported associations between cutaneous venous malformations and pigmented nevi. Further studies proposed that the vascular and pigmentary anomalies arises as a result of a genetic concept called twin spotting.[2,3] PPV was further delineated in five types with subtype 'a' for cutaneous involvement only and subtype 'b' for cutaneous as well as systemic involvement. Also, among five types of PPV, type II (Phakomatosis cesioflammea) was the most common with 75% reported cases.[4,5,6] However Goyal T et al[5] reported first case of Phakomatosis cesioflammea (typeIIb) from India in a 4-year old girl child. The largest series of PPV was published by Cordisco et al[7], who presented 25 patients in Argentina. In that, type IIb was the most common type. In another study it was reported that the most common association with extra cutaneous presentations was with the Sturge-Weber syndrome (SWS) and with the Klippel-trenaunay syndrome (KTS), individually or combined.[8] Okunola et al.[9] reported two cases of Phakomatosis pigmentovascularis type IIb in association with external hydrocephalus. Pradhan S et al[10] reported a case of Phakomatosis pigmentovascularis Type IIb with Sturge–Weber syndrome and cone shaped tongue. Jahangir et al.[11] reported a case of Phakomatosis pigmentovascularis with lower limb vascular abnormalities in a young Kashmiri male child.

Our patient had PWS, hypertrophy of left half of the face, trunk, extremities and external genitalia with venous engorgement on left lower abdomen and history of seizures. The color Doppler studies of lower limb showed chronic thrombosis of left deep venous system. MRI of the brain showed cerebral hemiatrophy on left side with loss of white matter more significant in temporo-parieto-occipital region. The case is being reported for its rarity and unusual presentation.

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