Sturge Weber Syndrome with Phakomatosis Pigmentovascularis: A case report

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INTRODUCTION

Sturge weber syndrome (SWS), or encephalotrigeminal angiomatosis is characterised by cutaneous angiomas (port wine stain), neurological abnormalities (leptomeningeal angiomas) and glaucoma.

Phakomatosis pigmentovascularis (PPV) is a congenital skin condition associated with cutaneous vascular malformation and pigmented lesions.

Traditional Classification[1]	New Classification[3]	Pattern of Nevus
Type I (a, b*)	–	Nevus flammeus + pigmented linear epidermal nevus
Type II (a, b*)	Cesioflammea	Nevus flammeus + blue spot ± naevus anaemicus
Type III (a, b*)	Spilorosea	Nevus flammeus + nevus spilus ± naevus anaemicus
Type IV(a, b*)	Cesiomarmorata	Nevus flammeus + dermal melanosis + nevus spilus ± naevus anaemicus
Type V(a, b*)	Unclassifiable	Cutis marmorata telangiectatica congenita + blue spot

*a: isolated cutaneous lesion, b: cutaneous and extracutaneous involvement.

Each type is further subdivided into 2 subtypes based on the absence (type a) or presence of extracutaneous involvement (type b).

OBJECTIVE

The co-existence of PPV and SWS is known but is rare. This report describes a case for its multi disciplinary team management of the complications falling in this diverse spectrum of phakomatosis.

CASE DESCRIPTION

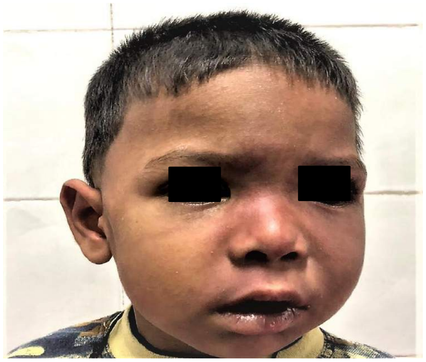
A 2-year-old developmentally normal boy born to non-consanguineous parents presented with recurrent episodes of right focal seizures and weakness of the right side of the body since 8 months of age.

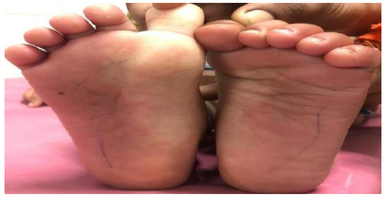
The child had a port wine stain on the left side of face and an extensive hyperpigmented lesion (Figure 1) involving the whole back extending till the trunk since birth. Pink hyperpigmentation was noted in soles of the feet (figure 2). Skin biopsy of the lesion at the back revealed to be a Mongolian spot (figure 4)

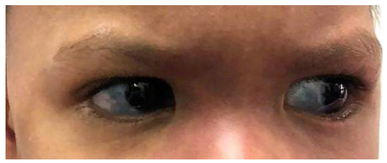
Eye examination showed bilateral blue pigmentation of the sclera (figure 3) with ocular melanosis. Subsequent ocular examinations revealed buphthalmos in the child's left eye.

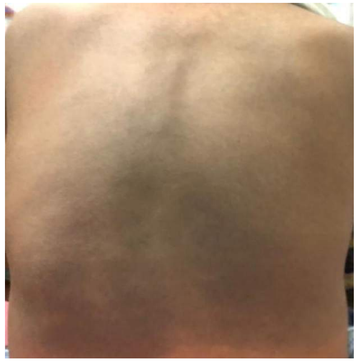
Neurological assessment showed right sided hemiparesis without any cranial nerve involvement with normal higher mental function. Neuroimaging showed diffuse left cerebral hemiatrophy with gyriform calcifications, prominent trans parenchymal veins and attenuated cortical branches of left MCA consistent with Sturge Weber Syndrome.

Blood samples sent for genetic testing did not yield any known mutation.









DISCUSSION

The pathogenesis of PPV is hypothesized to be due to dysplasia of the nerve cells and melanin cells of the embryonic neural crest.

SWS is distinguished by port wine stain which occurs unilaterally along dermatomes supplied by the ophthalmic and maxillary division of trigeminal nerve. Involvement of V1 area of the trigeminal area strongly reflect the presence of leptomeningeal angioma causing seizures whereas patients with V2 area involvement are most likely to develop glaucoma.

The aetiology of PPV is explained as a postzygotic mutation in the GNA11 or GNAQ gene. Sporadic somatic mutation in the GNAQ gene has been postulated to promote the RAS pathway stimulating proliferation and inhibition of apoptosis in people with SWS.

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