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Phakomatosis Pigmentovascularis Type II: Case Report

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We report a case of a 12 year old Asian girl. The patient presented to our OPD with congenital erythematous lesions on her face, trunk and both upper and lower extremities. There are large bluish-gray patches on trunk and extremities.

She was born full term through casearean section; weighed 3.1 kg and her mother experienced no pathological events during pregnancy and delivery. The family history was unremarkable for lesions. The general physical examination revealed bilateral naevus of ota with melanosis bulbi on her face and generalized port-wine stain (naevus flammeus) on her body. In addition, extensive bluish gray hyperpigmented patches (Mongolian spots) were located on the trunk and extemities. Developmental growth was achieved normally MRI of brain did not show abnormal findings. There is scoliosis detected both clinically and on lumbar spine x-ray. There is short 4th torso of right foot (Figures 1-3).

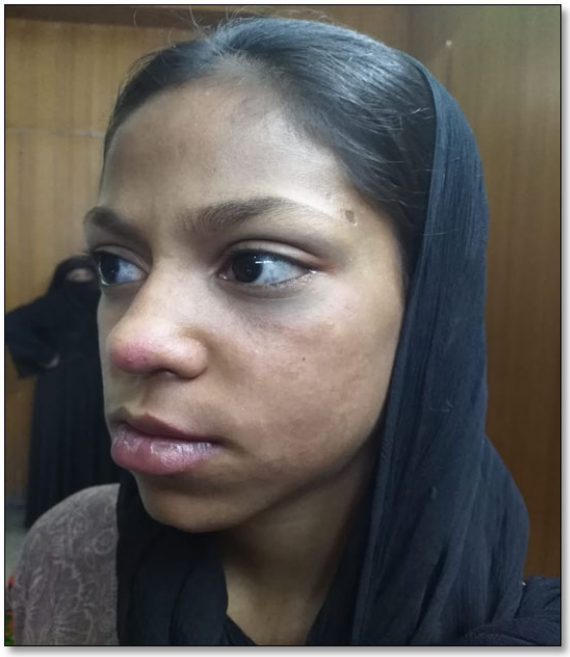

Figure 1. Bilateral naevus of ota with melanosis bulbi on her face and port-wine stain.

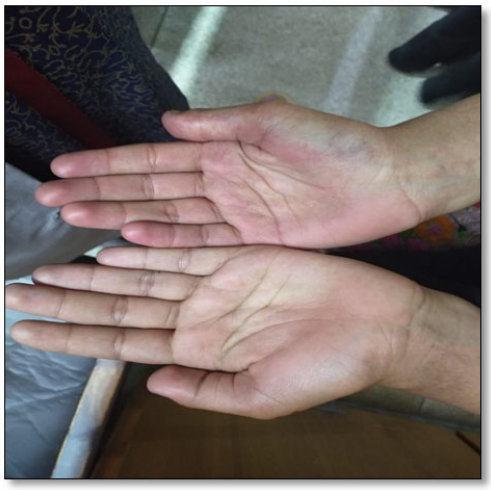

Figure 2. Port-wine stain on her palms.

In 1947, Ota et al. first described PPV as a disorder characterized by the combination of melanocytosis and capillary malformation [1]. Hasegama and Yasuhara classified PPV into four types according to combination of pigmentary skin lesions and naevus flammers [2]. In 2003 that is characterized by cutis marmorata telangiectatica [3,4]. Phacomatosis pigmento vascularis is defined as coexistence of widespreasd vascular naevus and extensive pigmentary naevus.

More than 250 sporadic cases of phacomatosis pigmentovascularis have been reported so far, mainly in Asians or Asian –related population [5]. Mutations in genes related to antigenic pathways (RAS, MAPK, mToR,

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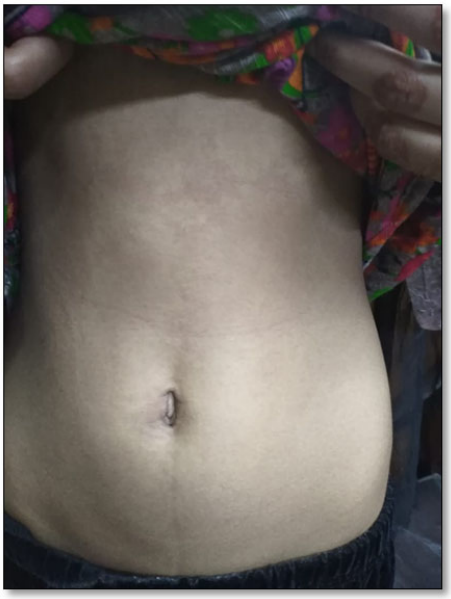

Figure 3. Bluish gray hyperpigmented patches on the trunk.

PI3K/AkT and GNAQ) have been recently identified as the causes of this complex phenotype [6]. In many cases, mutations in two different genes may coexist representing, the classic example of "twin spotting" phenomenon [7]. PPV is classified in four groups on the basis of phenotypes **(Table 1)**.

Ocular

Melanosis bulbi, glaucoma, iris mammillation, megalocornea buphthalmos, strabismus and hyperpigmentation of conjunctiva, sclera, episclera, iris, trabecular meshwork and choroid.

Musculoskeletal

Limb hypertrophy, klippel trenaunay type abmnormelitas, hemifacial hyphypertrophy, hemicorporal hypertrophy, macrocephaly microcephaly, scoliosis.

CNS

CNS anomalies include seizures, cognitive delay cerebral atrophy, hydrocephalus, sensory neural deafness, intracranial hypertension as well as migraine and intra-cerebral vascular malformations.

More rarely, PPV has associated with structural and or vascular renal anomalies hepato-splenomegaly, pyogenic granuloma cavernous haemangioma, portal hypertension umbilical hernia, hypoplasia of leg veins and hypo or hyperactivity of immune system.

Table 1. Classification of PPV.

Type of phacomatosis	Type of coexistant nevi	Traditional name	Additional skin lesions
Phacomatosis cesioflammea	Nevus cesius Nevus flammeus (port wine stain)	PPV type IIa/IIb	Nevus anemicus Areas of hairlessness Hypoplastic nails
Phacomatosis spilorosea	Nevus spilus (speckled lentiginous nevus) Telengiectatic nevus of the pale pink type	PPV type IIIa/IIIb	Granular layer tumors Areas of hairlessness Lymphoderma
Phacomatosis cesiomarmorata	Nevus (blue spot) Cutis marmorata telengiectatica congenita (CMTC)	PPV type Va/Vb	None
Unclassifiable type	Various types of vascular and pigmentary nevi, sometimes with overlapping phacomatosis cesioflammea and phacomatosis spilovascularis	PPV type IVa/IVb	CAL macules Hypomelanotic macules Nevus anemicus Nevus sebaceous

PPV is associated with several extra cutaneous anomalies.

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