



PAEDIATRIC DERMATOLOGY

PHACOMATOSIS PIGMENTOVASCULARIS: A RARE ENTITY

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Background: Phacomatosis Pigmentovascularis (PPV) is a heterogenous group of disorder, characterised by the co-presence of vascular malformation (most commonly naevus flammeus, also known as port wine stain) and pigmentary anomaly. They can occur with and without other systemic manifestations. Only approximately 250 cases have been described to date.

Observation(s): An otherwise well 10-year-old South-Asian female with no past medical history presented to the Dermatology Department with an eight month history of circular patches of depigmentation on her lower limbs bilaterally. She also had a capillary malformation on her right leg, noticed on day 2 of life, with an associated leg length discrepancy of 1.5cm. Furthermore, patchy areas of hyperpigmentation were noted on her left arm, left back and also on her bilateral lower limbs. After genetic testing showing a GNAQ mutation, a diagnosis of PPV was made. A second case of likely PPV will also be presented - genetic testing is in progress.

Key Message: PPV was first described by Ota in 1947. It is a rare condition, the genetic basis of which is still under investigation. However, investigators have found that it can be caused by a mosaic mutation in GNAQ and GNA11. Most classification systems describe 4 types of PPV, which can be further subclassified based on the presence or absence of systemic involvement. The most common systemic manifestations include skeletal, ocular, vascular and neurologic. Other manifestations include muscular and cardiac. Once PPV is suspected clinically, various investigations can be conducted to assess for systemic manifestations. As it has a variety of clinical presentations, it should be considered in any child with both a vascular and pigmentary abnormalities, with or without systemic manifestations. In patients without systemic involvement, treatment is not required. However, many children present to dermatology departments, seeking treatment due aesthetic concerns.

