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Case Report

Phacomatosis pigmentovascularis with Raynaud's phenomena

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Introduction

Phacomatosis pigmentovascularis is a rare disorder which occurs sporadically, with just about 200 cases reported worldwide.¹ The syndrome is characterized by a capillary nevus occurring in conjunction with a pigmented nevus of various types.² The condition may occur with systemic/syndromic involvement.³ The first reported case dates back to 1920, but it was only in 1947 that the condition was described in detail by Ota et al. Happle simplified and reclassified the disorder in 2005.⁴ We report a case of phacomatosis pigmentovascularis with Raynaud's phenomena in a young adult male.

Case report

A 27-year-old male patient presented with complaints of recurrent episodic left upper limb pain, swelling and bluish discoloration of fingers of left hand for six months duration. Symptoms aggravated on exposure to cold. Examination revealed the following skin lesions: (1) Multiple discrete to confluent, geographically shaped reddish-pink macular lesions (Port-wine stain) over the left anterior chest, crossing midline at the back, left arm, forearm and the left hand. [Fig. 1A and 1B], (2) In addition, he had a large, well-defined blue-black asymptomatic macular dermal pigmentary lesion over upper back and both upper limbs. On the back, both Port-wine stain and nevus fuscoceruleus were present independently, in close proximity to each other as well as superimposed on each other at places [Fig. 2]. All the lesions were present since birth and were stationary in nature vide history given by the patient, (3) Reddish-blue discoloration of ulnar half of left hand & ulnar four digits, with a positive Raynaud's test (cold stimulation test) [Fig. 3].

The hair, nails and mucus membranes were normal. All the limbs were symmetrical and only mild oedema of left hand was appreciated. No significant family history or history of consanguinity in parents was elicited. Neurologic examination was normal. Evaluation for aetiology of Raynaud's phenomenon including chest and cervical spine x-ray for cervical rib, CDFI (colour doppler flow index) left upper limb, MR (magnetic resonance) angiography for thoracic outlet syndrome and ANA screen for connective tissue disorder was non-contributory. He was referred to a higher tertiary care facility where he was managed with medical therapy in the

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Fig. 1 – Port-wine stain over the left anterior chest, left arm, forearm and the left hand. (A) Frontal aspect (B) Lateral aspect.

form of oral diltiazem and pregabalin and underwent left cervical sympathectomy, with partial symptomatic relief.

Discussion

The term phacomatosis actually refers to a developmental malformation simultaneously affecting tissues of ectodermal origin, namely eye, skin and central nervous system.² Presently, it is applied to few genetically determined diseases characterised by multiple nevi being present with or without systemic involvement. Phacomatosis pigmentovascularis is one such rare condition where a vascular nevus is present concomitantly with a pigmented nevus. The condition has been explained by the phenomenon of twin spotting which assumes that autosomal recessive mutations are present on

two different neighbouring loci and that they are exchanged simultaneously by somatic recombination.⁵

The first such case was reported in 1920 and in 1947 the condition was described in detail by Ota et al. Ruiz-Maldonado et al, in 1987 classified it into two basic subtypes i.e. Subtype A: Cutaneous forms and Subtype B: Systemic/syndromic forms.³ In 2005, Happle simplified and reclassified the condition into three types: phacomatosis cesioflammea, phacomatosis spilorosea and phacomatosis cesiomarmorata. He further dropped the term type I and proposed that the extremely rare unclassifiable form as type IV. Further, he proposed the term “phacomatosis multiplex” for some cases that could not be ascribed to any well-defined clinico-genetic entity.⁴ Phacomatosis cesioflammea is the most frequently occurring type. This type is characterized by a coexistence of one or more aberrant blue spots (Mongolian spots, dermal melanocytosis) and one or more Port-wine stains. The Latin word ‘caesius’

Fig. 2 – Port-wine stain and nevus fuscoceruleus co-existing on the back.

Fig. 3 – Reddish-blue discolouration of ulnar half & fingers of left hand.

means bluish gray and serves as an equivalent of the term fuscoceruleus to describe an aberrant Mongolian spot. The vascular nevus is usually purple-red and represents a nevus flammeus.

Both skin lesions may occur in isolation, or they may be associated with systemic involvement, be it in the form of central nervous system defects,³ ocular anomalies such as melanosis bulbi or glaucoma,^{6,7} asymmetrical length of limbs,^{8,9} dysplastic veins or lymph vessels^{6,10} or nevus anemicus.^{11,12} In our case, in addition to a dermal melanocytic nevus and a Port-wine stain, systemic involvement in the form of Raynaud's phenomenon was noted, which to the best of our knowledge has not been reported in literature till date.

Conflicts of interest

All authors have none to declare.

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