

Violaceous Patches on the Arm

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A 55-year-old man presented with red-violet patches on the right arm and chest that had been present since birth. The patches were asymptomatic and stable in size and shape. He denied any personal or family history of glaucoma or epilepsy. Physical examination demonstrated nonblanchable, violaceous to red patches on the right arm, back, and chest. No thrills or bruits were appreciable, and the right and left arms were of equal circumference and length. Further examination revealed hyperpigmented patches on the bilateral conjunctivae.

WHAT'S THE DIAGNOSIS?

- a. Klippel-Trenaunay syndrome
- b. phacomatosis cesioflammea
- c. phacomatosis cesiomarmorata
- d. Sturge-Weber syndrome
- e. tuberous sclerosis

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The authors report no conflict of interest.

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THE DIAGNOSIS: Phacomatosis Cesiioflammea

Phacomatosis pigmentovascularis (PPV) encompasses a group of diseases that have a vascular nevus coupled with a pigmented nevus.¹ It is divided into 5 types: Type I is defined by the presence of a vascular malformation and epidermal nevus; type II by a vascular malformation and dermal melanosis with or without nevus anemicus; type III by a vascular malformation and nevus spilus with or without nevus anemicus; type IV by a vascular malformation, dermal melanosis, and nevus spilus with or without nevus anemicus; and type V as cutis marmorata telangiectatica congenita and dermal melanosis.¹

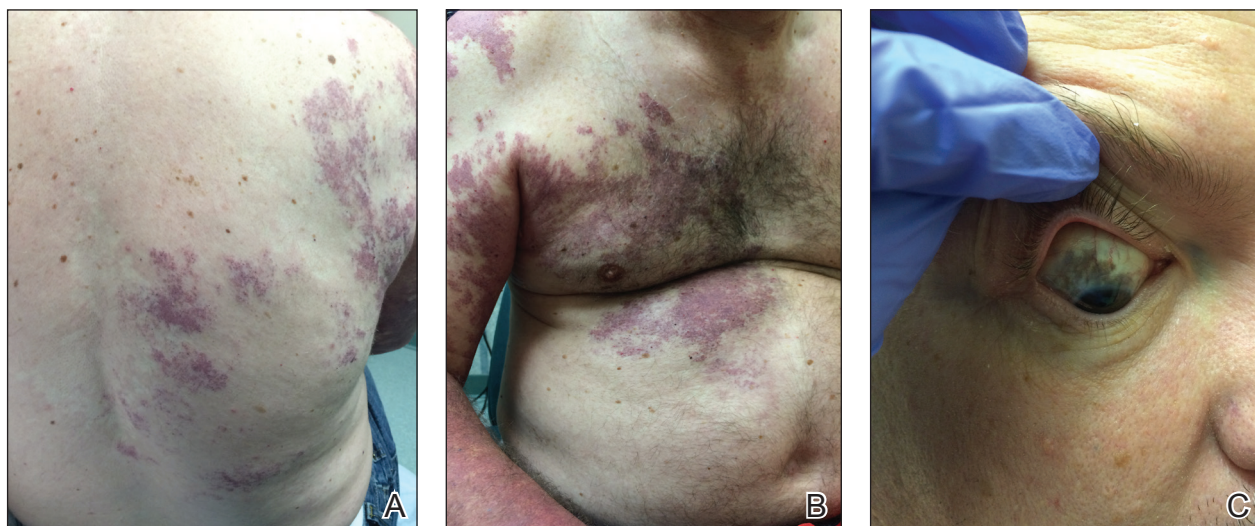
Happle² proposed a descriptive classification system in 2005 that eliminated type I PPV because neither linear epidermal nevus nor Becker nevus are derived from pigmentary cells. An appended “a” denotes a subtype with isolated cutaneous findings, while “b” is associated with extracutaneous manifestations. Phacomatosis cesiioflammea (type IIa/b) refers to blue-hued dermal melanocytosis and nevus flammeus. Phacomatosis spilorozea (type IIIa/b) refers to nevus spilus and rose-colored nevus flammeus. Phacomatosis cesiomarmorata (type Va/b) refers to dermal melanocytosis and cutis marmorata telangiectasia congenita. The last group (type IVa/b) is unclassifiable phacomatosis pigmentovascularis.^{2,3}

Phacomatosis pigmentovascularis can be isolated to the skin or have associated extracutaneous findings, including ocular melanocytosis, seizures, or cognitive delay due to intracerebral vascular malformations. Patients also can develop limb and soft-tissue overgrowth.⁴ Phacomatosis pigmentovascularis has been found to be associated

with mutations in the *GNA11* and *GNAQ* genes. The theory behind PPV is twin spotting, resulting from a somatic mutation that leads to mosaic proliferation of 2 different cell lines.⁵ Phacomatosis pigmentovascularis can occur in isolation or can demonstrate the phenotype of Sturge-Weber syndrome or Klippel-Trenaunay syndrome. In Sturge-Weber syndrome, capillary malformations involve the face and underlying leptomeninges and cerebral cortex. Glaucoma and epilepsy also may be present. In Klippel-Trenaunay syndrome, capillary malformations involve the extremities (usually the legs) in association with varicose veins, soft-tissue hypertrophy, and skeletal overgrowth.⁶⁻⁹ Tuberous sclerosis is an autosomal-dominant neurocutaneous disease in which patients develop hamartomas throughout the body, including the brain, skin, eyes, kidneys, heart, and lungs. Cutaneous manifestations include facial angiofibromas, ungual fibromas, hypomelanotic macules (ash leaf spots, confetti-like lesions), shagreen patches or connective tissue hamartomas, and fibrous plaques on the forehead. Tuberous sclerosis does not include vascular malformations.¹⁰

Our patient was diagnosed with PPV type IIb, or phacomatosis cesiioflammea. He had a large port-wine stain involving the right upper arm, back (Figure, A), and chest (Figure, B) with involvement of the bilateral conjunctivae (Figure, C). Our case is unique because our patient did not have dermal melanocytosis, only ocular melanocytosis.

Once underlying neurologic and vascular anomalies have been ruled out, port-wine stains can be treated cosmetically. Pulsed dye laser is the gold standard therapy



Phacomatosis cesiioflammea. A and B, Violaceous to red patches on the back and chest. C, Hyperpigmentation of the conjunctiva.

for capillary malformations, especially when instituted early. Follow-up with ophthalmology is advised to monitor ocular involvement. Shields et al¹¹ suggested dilated funduscopy for patients with port-wine stains because choroidal pigmentation often is the only ocular change seen. Ocular melanocytosis can progress to pigmented glaucoma or choroidal melanoma.

REFERENCES

1. Fernandez-Guarino M, Boixeda P, De las Heras E, et al. Phakomatosis pigmentovascularis: clinical findings in 15 patients and review of the literature. *J Am Acad Dermatol*. 2008;58:88-93.
2. Happle R. Phakomatosis pigmentovascularis revisited and reclassified. *Arch Dermatol*. 2005;141:385-388.
3. Villarreal DJ, Leal F. Phakomatosis pigmentovascularis of cesioflammea type. *An Bras Dermatol*. 2016;91(5 suppl 1):54-56.
4. Thomas AC, Zeng Z, Riviere JB, et al. Mosaic activating mutations in *GNA11* and *GNAQ* are associated with phakomatosis pigmentovascularis and extensive dermal melanocytosis. *J Invest Dermatol*. 2016;136:770-778.
5. Krema H, Simpson R, McGowan H. Choroidal melanoma in phakomatosis pigmentovascularis cesioflammea. *Can J Ophthalmol*. 2013;48:E41-E42.
6. Wu CY, Chen PH, Chen GS. Phakomatosis cesioflammea associated with pectus excavatum. *Acta Derm Venereol*. 2009;89:301-310.
7. Pradhan S, Patnaik S, Padhi T, et al. Phakomatosis pigmentovascularis type IIb, Sturge-Weber syndrome and cone shaped tongue: an unusual association. *Indian J Dermatol Venereol Leprol*. 2015;81:614-616.
8. Turk BG, Turkmen M, Tuna A, et al. Phakomatosis pigmentovascularis type IIb associated with Klippel-Trenaunay syndrome and congenital triangular alopecia. *J Am Acad Dermatol*. 2011;65:E46-E49.
9. Sen S, Bala S, Halder C, et al. Phakomatosis pigmentovascularis presenting with Sturge-Weber syndrome and Klippel-Trenaunay syndrome. *Indian J Dermatol*. 2015;60:77-79.
10. Schwartz RA, Fernandez G, Kotulska K, et al. Tuberous sclerosis complex: advances in diagnosis, genetics, and management. *J Am Acad Dermatol*. 2007;57:189-202.
11. Shields CL, Kligman BE, Suriano M, et al. Phakomatosis pigmentovascularis of cesioflammea type in 7 patients: combination of ocular pigmentation (melanocytosis or melanosis) and nevus flammeus with risk for melanoma. *Arch Ophthalmol*. 2011;129:746-750.