CASE REPORT

Phakomatosis pigmentovascularis type IIb: A case report.

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Key words: phakomatosis pigmentovascularis; aberrant Mongolian spots; port-wine stain; genetic mosaicism; GNA11; GNAQ

How to cite this article:
Rosero J., Narváez C., Rosero E. Facomatosis pigmentovascular tipo IIb: reporte de caso clínico. - Rev Dermatol Cent Úraga. 2026;8(2).

Date of receipt: 3/3/2026
Date of acceptance: 30/7/2026

ABSTRACT

Phakomatosis pigmentovascularis (PPV) is a rare genodermatosis (estimated prevalence of 5.8 per 100,000 pediatric patients) characterized by the coexistence of capillary vascular malformations and dermal melanocytosis, resulting from postzygotic somatic mosaic mutations in the GNA11 and GNAQ genes. We report the case of a 3-year-old girl presenting with aberrant Mongolian spots, a port-wine stain, and nevus anemicus since birth, associated with expressive language delay, consistent with type IIb phakomatosis pigmentovascularis (phakomatosis cesioflammea with extracutaneous involvement). A multidisciplinary follow-up program was initiated, and brain magnetic resonance imaging is pending. This case contributes to the limited literature on this uncommon disorder from Latin America.

INTRODUCTION

Phakomatosis pigmentovascularis (PPV) is a rare congenital syndrome characterized by the coexistence of an extensive capillary vascular malformation (port-wine stain) and a pigmented lesion derived from dermal melanocytes. First described by Ota in 1947, PPV was reclassified by Happle in 2005 into four descriptive groups, with phakomatosis cesioflammea (aberrant Mongolian spots associated with a port-wine stain) representing the most common subtype. Each group is further subdivided into subtype "a," limited to cutaneous involvement, and subtype "b," which includes extracutaneous manifestations.[1,2]

At the molecular level, PPV is associated with postzygotic activating somatic mosaic mutations in the GNA11 (p.Arg183Cys) and GNAQ (p.Arg183Gln) genes, which encode Gα subunits of heterotrimeric G proteins involved in the activation of the MAPK signaling pathways, including p38, JNK, and ERK.[3] These mutations are detectable at low variant allele frequencies (1.5%–8.6%) in affected tissues but are absent in peripheral blood, confirming their postzygotic mosaic nature.[3,4] Although PPV generally follows a benign clinical course, it may be associated with neurological abnormalities (including hydrocephalus and seizures), ophthalmologic complications (particularly glaucoma, reported in up to 88% of systematically evaluated eyes), and musculoskeletal abnormalities.[2,5,6]

CASE REPORT

A 3-year-old girl, the first child of a healthy pregnancy, presented with no significant prenatal or perinatal history. Her past medical history was notable for atopic dermatitis and allergic rhinitis. Her parents sought medical evaluation because of progressively

enlarging hyperpigmented lesions over the lumbar and gluteal regions that had been present since birth.

Physical examination: Multiple blue-gray macules with irregular, poorly defined borders, measuring 2–8 cm in diameter, were observed over the bilateral lumbar, gluteal, and sacral regions, consistent with aberrant Mongolian spots. A 4 × 3 cm pink-to-erythematous macule was present on the right paravertebral lumbar region, consistent with a port-wine stain. A 3 × 2 cm area of relative hypopigmentation was identified on the left lumbar region, compatible with nevus anemicus. Neurological examination revealed expressive language delay without additional focal neurological deficits.

Diagnosis: Type IIb phakomatosis pigmentovascularis (phakomatosis cesioflammea with extracutaneous involvement) was diagnosed based on the coexistence of aberrant Mongolian spots, a port-wine stain, and nevus anemicus, with the expressive language delay considered a potential extracutaneous manifestation. Brain magnetic resonance imaging (MRI) was requested and was pending at the time of this report. The patient was enrolled in multidisciplinary follow-up involving dermatology, neurology, medical genetics, and ophthalmology.

DISCUSSION

Phakomatosis pigmentovascularis (PPV) is a neurocutaneous syndrome belonging to the spectrum of disorders caused by mosaic mutations affecting heterotrimeric G proteins, together with Sturge–Weber syndrome and McCune–Albright syndrome.[3,7] Phakomatosis cesioflammea is the most common subtype, as demonstrated in the case series reported by Vidaurri-de la Cruz (24 patients), Fernández-Guarino (15 patients), and Shin (52 patients).[2,8,9] In 2016, Thomas et al. identified activating GNA11 and GNAQ mutations using deep sequencing of affected tissue, while zebrafish models successfully recapitulated the dermal melanocytosis observed in humans.[3] More recently, a Chinese cohort of 13 pediatric patients confirmed GNA11/GNAQ mosaicism in 92.3% of cases, and Sliepka et al. identified a GNA11 mutation directly in brain tissue, providing evidence that mosaicism may also involve the central nervous system (CNS).[4,10]

The principal differential diagnoses include isolated extensive Mongolian spots (without an associated vascular component), Sturge–Weber syndrome (facial capillary malformation associated with ipsilateral leptomeningeal



Figure 1. Aberrant Mongolian spots involving the lumbosacral region, a right paravertebral port-wine stain, and a right lumbar nevus anemicus.

angiomatosis, without dermal melanocytosis), Klippel–Trénaunay syndrome (capillary malformation associated with limb hypertrophy and venous and/or lymphatic malformations), isolated nevus of Ota or nevus of Ito, and cutis marmorata telangiectatica congenita. The hallmark diagnostic feature of PPV is the simultaneous coexistence of dermal melanocytosis and a congenital capillary vascular malformation.[1,11] In our patient, the combination of aberrant Mongolian spots, a port-wine stain, and nevus anemicus established the diagnosis unequivocally.

Systemic evaluation is mandatory in patients with PPV. Neuroimaging should be performed in the presence of any neurological manifestations, ophthalmologic assessment should include tonometry because glaucoma is the most frequent ocular complication,[6] and vascular imaging should be considered in patients presenting with limb asymmetry.[2,8] Okunola et al. reported two cases of type IIb PPV associated with hydrocephalus, emphasizing the importance of comprehensive neurological evaluation even in patients who appear clinically asymptomatic.[5] More recent reports further support individualized neurological, ophthalmologic, and vascular surveillance beginning in early childhood.[12]

Treatment is primarily symptomatic. Pulsed dye laser (595 nm) is the treatment of choice for port-wine stains, whereas Q-switched Nd:YAG laser (1064 nm) or Q-switched alexandrite laser (755 nm) may improve dermal melanocytosis, although cosmetic outcomes are variable.[9,13,14] Early treatment during childhood is recommended to optimize therapeutic results.[15] Prognosis is generally favorable in patients with isolated cutaneous disease, whereas outcomes in those with extracutaneous involvement depend largely on the severity of associated neurological and ophthalmologic complications.[5,6,10]

CONCLUSIONS

Type IIb phakomatosis pigmentovascularis is an uncommon neurocutaneous syndrome whose early clinical recognition—based on the coexistence of congenital dermal melanocytosis and a capillary vascular malformation—allows timely surveillance for potentially serious extracutaneous complications. The identification of GNA11 and GNAQ mutations has clarified the molecular pathogenesis of this disorder and established its place within the spectrum of mosaic G-protein disorders. Reporting this case from Ecuador contributes to the limited evidence available from underrepresented Latin American populations.

INFORMED CONSENT

Written informed consent for the use of the patient's clinical data and images for research and scientific publication was obtained from the patient's legal guardians. Patient confidentiality was maintained, and no identifying information has been disclosed.

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