ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.1.2026.25.32

Journal of Dermatological Case Reports

Phakomatosis Pigmentovascularis: A Case Report and Review of Clinical Subtypes

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Abstract:

Background: Phakomatosis pigmentovascularis (PPV) is a rare congenital neurocutaneous disorder defined by the coexistence of pigmentary lesions and vascular malformations. PPV type II (phakomatosis cesioflammea) is the most common subtype and is characterized by dermal melanocytosis and capillary malformations. Early recognition is important due to potential ocular, neurologic, and systemic involvement.
Case Presentation: We report a newborn female presenting with extensive slate-gray to blue dermal melanocytosis involving the back, extremities, and abdomen, along with bilateral facial violaceous capillary malformations. Systemic evaluation, including abdominal ultrasonography and brain MRI with angiography, revealed no visceral or intracranial vascular anomalies. Laboratory studies were unremarkable aside from hemoglobin S trait. The cutaneous findings were consistent with PPV type II. Pediatric neurology, dermatology, and ophthalmology referrals were arranged to evaluate for associated glaucoma, neurologic abnormalities, and to initiate long-term surveillance. Future pulsed-dye and Nd:YAG laser therapy was recommended for lesion management.
Discussion: PPV is extremely rare, with fewer than 200 cases documented. The condition may phenotypically overlap with Sturge–Weber syndrome, Klippel–Trenaunay syndrome, and other vascular anomalies, necessitating careful systemic evaluation. Although the pathogenesis is linked to somatic mosaic mutations affecting RAS/MAPK signaling, clinical diagnosis remains paramount. Early multidisciplinary management is essential to monitor for ocular and neurologic complications and to optimize cosmetic outcomes.
Conclusion: This case highlights the characteristic presentation of PPV type II and reinforces the importance of comprehensive systemic evaluation and multidisciplinary follow-up. Awareness of this rare entity aids in distinguishing it from other neurocutaneous syndromes with overlapping features.

Keywords:

Phakomatosis pigmentovascularis, nevus flammeus, congenital dermal melanocytosis, capillary malformation

Received : 28-12-2025 **Revised : 21-01-2026** **Accepted: 21-01-2026** **Published : 28-01-2026**

Introduction

Phakomatosis pigmentovascularis (PPV) is a rare congenital neurocutaneous syndrome characterized by the coexistence of a pigmentary nevus and a vascular malformation.[1] The condition is classified into several subtypes based on the specific combination of pigmentary and vascular lesions, with phakomatosis cesioflammea (either nevus of Ota or Mongolian spot with port-wine

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stain) being the most common.[2] Nevi of Ota are benign dermal melanocytic lesions presenting as blue-gray or brown hyperpigmentation along the V1 and V2 trigeminal distributions, most often affecting the periocular skin, temple, forehead, and sometimes ocular or other mucosal sites.[3] A related form of dermal melanocytosis is the Mongolian spot, which presents as blue-gray to greenish macules most often on the lumbosacral and gluteal regions of newborns.[4] In contrast, A port-wine stain (nevus flammeus) is a congenital capillary malformation presenting at birth as a well-demarcated pink to red patch, most often on the face and neck, that progressively darkens to red-purple, and unlike other vascular birthmarks, persists throughout life without spontaneous regression.[5]

Systemic involvement in PPV is variable and may include ocular abnormalities (notably glaucoma and choroidal alterations), neurologic manifestations (such as epilepsy or seizures), and overgrowth or asymmetry of limbs.[6] Ocular involvement is frequent and can predispose to complications such as glaucoma and, rarely, uveal melanoma.[7] The risk of systemic complications is higher when pigmentary lesions are located on the head and neck or when nevus of Ota is present.[1] Diagnosis is clinical, based on recognition of the characteristic skin findings, with further evaluation for systemic involvement as indicated.[8] We now describe a patient with features of phakomatosis pigmentovascularis and possible Sturge-Weber syndrome, illustrating the clinical overlap and diagnostic challenges of these conditions.

Patient Presentation

A newborn female infant was noted at delivery to have extensive cutaneous lesions. The infant was born at 39w3d via spontaneous vaginal delivery to a 28-year-old G2P2 mother of Filipina descent with a negative prenatal infectious disease panel, A+ blood type, and no history of pregnancy complications. The father is of Costa Rican descent and is a sickle cell disease carrier. The infant's APGAR scores were 8 at 1 minute and 9 and 5 minutes. Weight was 3.171 kg, length was 19 in, and head circumference was 31.5 cm. Physical exam was unremarkable with the exception of diffuse, slate-gray to blue macular pigmented lesions prominent over the entirety of the back, extending continuously from the lumbosacral region and buttocks to the upper thoracic area. Additional patches were observed on both upper and lower extremities, with partial involvement of the anterior abdomen. The borders were ill-defined, and the distribution was asymmetric. No associated elevation, induration, or tenderness was noted. Examination also revealed violaceous patches with well-defined borders on both cheeks, more pronounced on the left side, consistent with capillary vascular malformations. The overlying skin was intact without nodularity, ulceration, or tenderness.

Given the extent of cutaneous vascular malformations, abdominal ultrasonography was performed to evaluate for visceral involvement. The liver, kidneys, and spleen were normal in size and echotexture, with no evidence of vascular malformations. Brain MRI with angiography was obtained to assess for intracranial vascular abnormalities and was within normal limits. Laboratory studies, including complete blood count and comprehensive metabolic panel, were unremarkable. The newborn metabolic blood screen revealed no abnormalities, except for a hemoglobin S trait. No additional laboratory abnormalities were identified.

The constellation of pigmentary and vascular findings was most consistent with phakomatosis pigmentovascularis (PPV), a rare neurocutaneous syndrome characterized by the association of pigmentary and vascular anomalies. Based on the clinical findings, this presentation aligns with PPV type II. This type of PPV involves the presence of dermal melanoctyosis in conjunction with capillary malformations.

The infant remained clinically stable throughout hospitalization and was discharged in good condition on hospital day two. A referral to pediatric neurology was arranged for further evaluation and surveillance. In addition, the patient was referred to ophthalmology to rule out associated congenital glaucoma. Pediatric

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dermatology was consulted during the admission and recommended outpatient follow up, noting that the infant would likely be an appropriate candidate for future pulsed dye laser therapy.

Discussion

In 1947, the term phakomatosis pigmentovascularis (PPV) was introduced to describe the association of extensive, atypical, and persistent nevus flammeus with pigmentary abnormalities.[9] PPV is an extremely rare condition, with institutional data reporting frequencies of approximately 5.8 per 100,000 pediatric patients and 0.63 per 100,000 dermatology patients; however, the true population prevalence remains unknown due to underreporting and variability in diagnosis.[10] Recent multicenter cohorts confirm its rarity, as well as its male predominance.[2] To date, only about 200 cases have been reported in the medical literature, with the majority documented in patients of Japanese origin.[11] The diagnosis is made clinically based on characteristic cutaneous findings.[1] Systemic evaluation is warranted to detect associated neurologic, vascular, or other organ involvement, since significant syndromic associations can mimic the findings seen in PPV.[9,12]

The most widely accepted classification is the Happle system, which recognizes four subtypes. Phakomatosis cesioflammea, or Type II, is defined by the coexistence of a nevus flammeus and dermal melanocytosis, which includes Mongolian spots and nevus of Ota.[8] This is the most common subtype of PPV.[2] Phakomatosis spilorosea, or Type III, is characterized by the presence of a nevus spilus along with a capillary malformation, and has been associated with PTPN11 mosaicism.[13] A nevus spilus is a congenital melanocytic lesion characterized by a light brown background macule with superimposed darker speckles that represent melanocytes, which may appear as either flat macules or raised papules.[14] Phakomatosis cesiomarmorata, or Type IV, is defined by the coexistence of dermal melanocytosis and cutis marmorata telangiectatica congenita (CMTC).[6] CMTC is a rare congenital capillary malformation presenting as persistent, reticulated, marbled erythema of the skin, often with associated cutaneous atrophy, ulcerations, and body asymmetry.[15] The final subtype is a group of rare combinations that do not fall into any of the previous categories.[8]

Recent genetic studies have identified somatic mosaic mutations in genes such as GNAQ, GNA11, and PTPN11 as causative in various PPV subtypes, supporting a mosaic pathogenesis driven by dysregulation of the RAS/MAPK signaling pathway.[13] Mutations in these pathways appear to cause aberrant development of both vascular and melanocytic tissues.[13] Ocular manifestations are common and may include glaucoma, choroidal hemangiomas, and pigmentary changes that increase the risk of uveal melanoma.[7] Systemic features may include epilepsy, anemia, limb asymmetry, and CNS anomalies.[2,6]

Management of PPV is multidisciplinary and tailored to specific manifestations. One study showed that capillary malformations were nearly cleared in 28.6% of patients with pulsed dye laser, while pigmented nevi were nearly or completely cleared in 65.8% with Q-switched Nd:YAG laser.[1] It is preferred to initiate combined laser treatment for PPV in childhood under general anesthesia, as it reduces the number of sessions, improves quality of life, and increases cost-effectiveness.[16] Pigmented nevi responded better to laser therapy than capillary malformations, with smaller lesions showing superior outcomes in both.[1] Regular ophthalmic surveillance is essential due to risk of late-onset glaucoma and uveal melanoma. [7,17] Despite the varied mechanisms of glaucomatous damage, management parallels that of congenital and primary open-angle glaucoma, with topical intraocular pressure-lowering drops as first-line therapy.[18] Surgical options such as goniotomy, trabeculotomy, trabeculectomy, or tube shunt placement may be necessary in more severe cases.[2,18] Early and periodic multidisciplinary evaluation is recommended to monitor for progression and complications, especially ocular and neurologic sequelae.[2]

Appropriate consideration of differential diagnoses is critical for patients with PPV, as it can closely mimic multiple serious conditions. Its presentation

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can closely resemble Sturge-Weber syndrome, which is marked by facial capillary malformations, leptomeningeal involvement of the brain, glaucoma, and seizures.[19] Another important differential is Klippel-Trenauny Syndrome, which is characterized by venous malformation, port-wine stain, and limb hypertrophy.[20] Blue rubber bleb nevus syndrome is rare condition defined by multiple venous malformations and hemangiomas affecting the skin and visceral organs.[21] The most common manifestations are gastrointestinal bleeding and secondary iron deficiency anemia, but rupture, intestinal torsion, or intussusception may occur, which can be life-threatening.[21] The possibility of one of these underlying conditions presenting similarly to PPV necessitates systemic evaluation to rule them out or begin necessary treatment early.

Conclusion

This case highlights the classic presentation of phakomatosis pigmentovascularis type II in a newborn and underscores the diagnostic challenges posed by its clinical overlap with other neurocutaneous and vascular syndromes. Although initial systemic evaluation revealed no extracutaneous involvement, ongoing multidisciplinary follow-up remains essential given the potential for delayed-onset ocular and neurologic complications. Early identification and appropriate surveillance allow for timely intervention, optimize visual and developmental outcomes, and provide families with anticipatory guidance regarding prognosis and therapeutic options such as laser treatment. Continued reporting of cases like this contributes to a better understanding of the phenotypic spectrum, natural history, and management considerations of this rare disorder.

Figure 1.

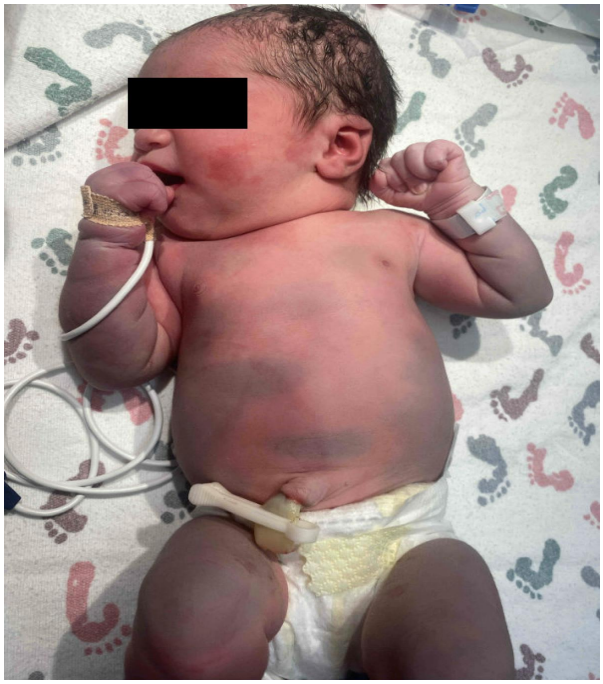

Figure 1 shows the extensive congenital dermal melanocytosis visible on the trunk and limbs, alongside facial capillary malformations.

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Figure 2

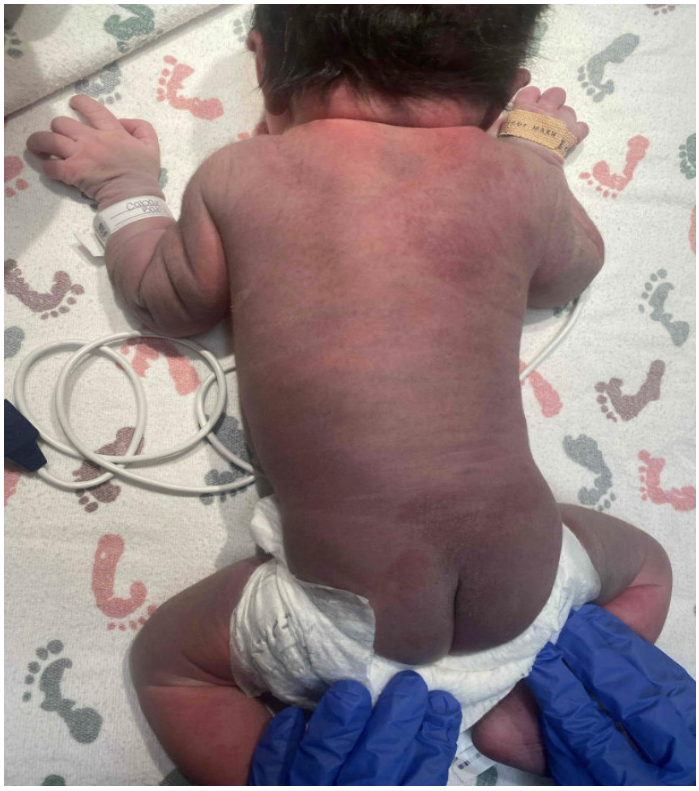

Figure 2 demonstrates widespread congenital dermal melanocytosis involving the back, trunk, and extremities of the infant.

Figure 3

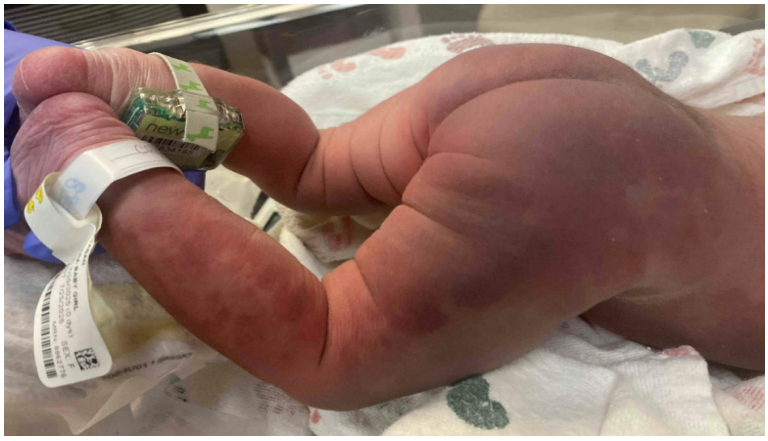

Figure 3 depicts a lateral view of the infant demonstrating congenital dermal melanocytosis involving the buttocks and lower extremities.

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Figure 4

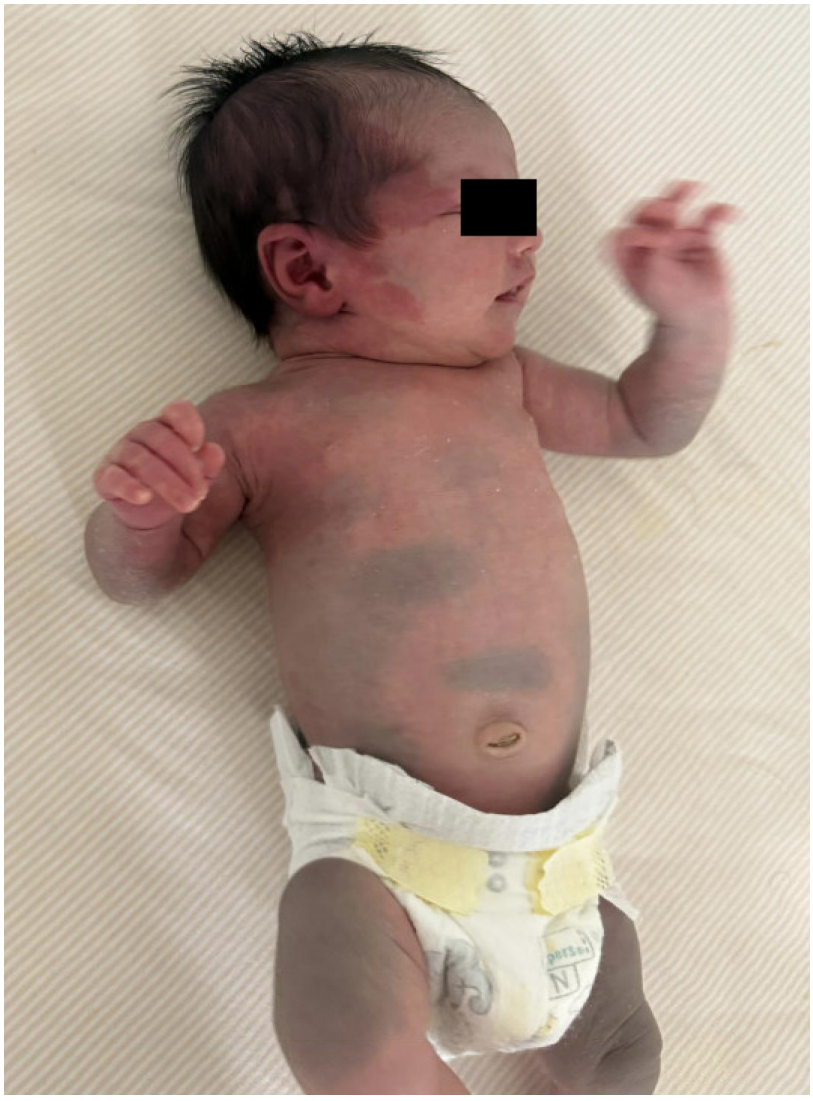

Figure 4 shows an anterior view of the infant showing congenital dermal melanocytosis on the trunk and limbs, with the cheek turned to reveal more extensive facial capillary malformation

Table 1 – CBC

Test	Reference Range	Result
WBC	7.5 - 15.8 10^3/uL	18.7
RBC	3.79 - 4.76 10^6/uL	5.19
Hemoglobin	12.7 - 16.4 g/dL	18.8
Hematocrit	36.5 - 47.7 %	54.2
MCV	89.7 - 105.4 fL	104.5
MCH	31 – 37 pg	36.2
MCHC	31.7 - 36.3 g/dL	34.7

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RDW	15 – 20%	16.6
Platelets	133 - 255 10^3/uL	285
Neutrophils%	21.0 - 55.0 %	68.3
Lymphocytes%	11.0 - 40.0 %	20.7
Monocytes%	5.0 - 11.0 %	7.9
Eosinophils%	2.0 - 4.0 %	2.3
Basophils%	0.0 - 1.0 %	0.8
ANC (auto diff)	4.43 - 11.43 10^3/uL	12.80
Lymphocytes Absolute	1.68 - 2.85 10^3/uL	3.90
Monocytes Absolute	0.57 - 1.72 10^3/uL	1.50
Eosinophils Absolute	0.05 - 0.32 10^3/uL	0.40
Basophils Absolute	0.02 - 0.07 10^3/uL	0.20

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