International Journal of Medical Science and Clinical Research Studies

ISSN(print): 2767-8326, ISSN(online): 2767-8342

Volume 05 Issue 03 March 2025

Page No: 475-477

DOI: https://doi.org/10.47191/ijmscrs/v5-i03-11, Impact Factor: 8.188

# Phacomatosis Pigmentovascularis: Genetic Insights through Cutaneous Pigmentation

**Veronica Martinez Garcia[1], Lucia Achell Nava[1], Guadalupe Maldonado Colin[1], Dolores Maribel Arellano Vivero[1], Carla Fernandez Reynoso[1]**

[1]Centro Medico Nacional 20 de Noviembre. Instituto de Seguridad y Servicios Sociales de los Trabajadores del Estado. Felix Cuevas 540. Colonia del Valle Sur. Benito Juarez. 03104. Ciudad de Mexico, Mexico.

**ABSTRACT**

Phacomatosis pigmentovascularis is a group of alterations involving a vascular malformation and a melanocytic lesion, and have diverse clinical cutaneous manifestations, which is the base for the current classification. Furthermore, systemic alterations can occur, including ophthalmological and neurological alterations, some with long term sequalae potential. The case of a 16-day old male patient without previous illness presenting with vascular and pigmented lesions is presented here, with subsequent referral to rule out the most important systemic alterations.

**KEYWORDS:** Phacomatosis pigmentovascularis, Phacomatosis, Capillary malformations, Neurocutaneous disorders, Congenital syndromes, Vascular anomalies

**ARTICLE DETAILS**

**Published On:**
**11 March 2025**

**Available on:**
**https://ijmscr.org/**

## I. INTRODUCTION

Phacomatosis pigmentovascularis refers to the coexistence of a vascular malformation and a melanocytic lesion (1). This is a group of rare diseases that may occur solely on the skin or be accompanied by systemic disorders (2).

## II. CASE PRESENTATION

A 16-day-old male patient was brought for assessment because of pigmented and red lesions reported by his mother since birth. The physical examination showed disseminated dermatosis to the face that covers the left half of it, and the anterior and posterior thorax in the left half consisting of a vascular aspect patch with segmental distribution and cross-linked pattern of irregular shape and size. This lesion was superimposed with another one located in the posterior thorax in the lumbar area characterized by a dark gray patch of irregular shape and size with well-defined borders. (Figure 1)

### *A. Course and outcome*

The diagnosis of phacomatosis pigmentovascularis is established and reference is made to the following specialties: ophthalmology, where the possibility of glaucoma is ruled out; neurology for complete examination, where no alterations are reported, in addition to genetics, without relevant findings. The major associated systemic alterations were ruled out.

Follow-up appointments and general skin care measures were granted.

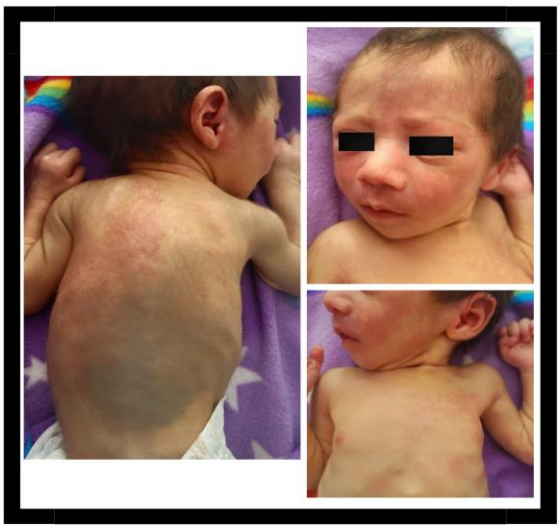

**Figure 1. Phacomatosis pigmentovascularis, subtype cesioflammea. A pigmentary lesion is observed in the posterior trunk that overlaps with a vascular lesion in the face, plus the posterior and anterior trunk.**

## III.DISCUSSION

Phacomatosis pigmentovascularis encompasses two different types of skin lesions, and its early detection by possible associations or extracutaneous manifestations is important; the most relevant aspects of this condition are discussed below.

### *A. Pathogenesis*

The cause is not well established; however, an association has been found between phacomatosis and the mutation in GNAQ (Nucleotide Binding Protein Guanine, Polypeptide Q) (2). A "twin-spotting" phenomenon, also called didymosis, has also been proposed in which somatic mutations occur in adjacent genes, causing mosaic spots very close to each other. (3) (4)

### *B. Classification*

It was first described in 1947 by Ota, its first classification included 5 subtypes, adding "a" for isolated lesions and "b" for extracutaneous lesions. (4):

Type I: Capillary malformation associated with epidermal nevus.

Type II: Capillary malformation associated with dermal melanosis, with or without anemic nevus.

Type III: Capillary malformation associated with spilus nevus, with or without anemic nevus.

Type IV: Capillary malformation, dermal melanosis, and spilus nevus, with or without anemic nevus.

Type V: Cutis marmorata telangiectasia congenital associated with dermal melanosis.

A new classification by Happle was later proposed, with four subtypes (4):

1. Phacomatosis cesioflammea (type IIa and IIb)
2. Phacomatosis spilorosea (type IIIa and IIIb)
3. Unclassifiable forms of phacomatosis (type IV)
4. Phacomatosis cesiomarmorata (type V)

Of the aforementioned, phacomatosis cesioflammea has been reported as the most frequent in 85%, within this subtype there is a greater tendency to extracutaneous manifestations and greater severity, whereas the spilorosea variety has been associated with multiple granular cell tumors. (5) (1). Phacomatosis cesiomarmorata or type V is the second most frequent. (6)

### *C. Extracutaneous manifestations*

Several extracutaneous manifestations, associated with phacomatosis pigmentovascularis, have been documented, occur in 50% of cases (5), and include: ocular melanocytosis or episcleral hyperpigmentation (the most common), glaucoma (the second most frequent), extremity hemihypertrophy, convulsions, ocular melanoma, bilateral deafness, and malignant hypertension. (2) (5) (1). Ocular manifestations are the most frequent and usually occur ipsilateral to a facial capillary lesion. (2) (5).

Associations: Phacomatosis cesioflammea subtype may be associated with syndromes such as Sturge-Weber. (4)

### *D. Management*

Management depends on the existence or absence of systemic manifestations, when the clinical expression of phacomatosis pigmentovascularis is purely cutaneous it presents with a benign course and does not require treatment, except for aesthetic reasons. However, when systemic manifestations exist, they should be addressed in a targeted manner (4).

For pigmentary and/or vascular lesions that impact the patient's quality of life due to their aesthetic perception, treatments such as pulsed light or Q-switch laser can be offered, the first being most useful for pigmentary lesions and the second for vascular lesions. (4)

## CONCLUSIONS

We present the case of a patient with phacomatosis pigmentovascularis, classified as IIa (cesioflammea), in whom the most frequent systemic alterations were ruled out by ophthalmology and neurology. The present case shows

the relevance of early detection to achieve a major functional prognosis.

The ocular and neurological assessment is particularly relevant, for its potential to cause irreversible sequelae such as permanent blindness, it is therefore appropriate to carry out the referral to these specialties early. Neurological and ophthalmological explorations are mandatory to rule out extracutaneous associations.

**REFERENCES**


I. James, W. D., Berger, T., & Elston, D. (2025). Andrew's diseases of the skin. Elsevier Health Sciences. pp. 635

II. Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. R. Z., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Undiagnosed Diseases Network, Mackenzie, K. M., Eng, C. M., Fisher, P. G., Ashley, E. A., Teng, J. M., Stevenson, D. A., Shieh, J. T., Wheeler, M. T., … Bernstein, J. A. (2019). Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. American journal of medical genetics. Part A, 179(6), 966–977. https://doi.org/10.1002/ajmg.a.61134Kola

III. Kola, B., Mehrafza, M., & Kane, A. (2019). Phacomatosis Pigmentovascularis: Simple Presentation of a Not So Simple Dermatological Condition. Clinical pediatrics, 58(4), 485–487. https://doi.org/10.1177/0009922817737088

IV. Villarreal, D. J., & Leal, F. (2016). Phacomatosis pigmentovascularis of cesioflammea type. Anais brasileiros de dermatologia, 91(5 suppl 1), 54–56. https://doi.org/10.1590/abd1806-4841.20164516

V. Huang, S., Yang, Y., Ouyang, Z., Guo, X., Zhan, Z., Liu, X., Chen, Z., Xu, J., & Yu, M. (2022). The risk of glaucoma associated with phacomatosis cesioflammea and phacomatosis cesioflammeo-marmorata. The risk of glaucoma associated with phacomatosis cesioflammea and phacomatosis cesioflammeo-marmorata. European journal of dermatology : EJD, 32(5), 589–596. https://doi.org/10.1684/ejd.2022.4317

VI. Torchia D. (2021). Phacomatosis spilorosea versus phacomatosis melanorosea: a critical reappraisal of the worldwide literature with updated classification of phacomatosis pigmentovascularis. Acta dermatovenerologica Alpina, Pannonica, et Adriatica, 30(1), 27–30.