Beyond an Extensive Dermal Melanocytosis: A Case of Phacomatosis Pigmentovascularis of Cesioflammea Type

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Keypoints

What is known:

- Dermal melanocytosis, also known as Mongolian spot, is a benign diagnosis in newborns, while it is extensive in certain ethnic groups.
- In rare situations, dermal melanocytosis can be associated with systemic diseases.

What is added:

- Phacomatosis pigmentovascularis should be considered in the differential diagnosis of congenital dermal disorders, namely aberrant dermal melanocytosis.
- In the absence of extracutaneous manifestations, phacomatosis pigmentovascularis of cesioflammea type is a benign condition. However, laser treatments can be used to improve the quality of life of patients.

Introduction

This study aimed to present the case of a 2-day-old male newborn with an extensive congenital dermal disorder. Both the pregnancy and family history were unremarkable. He presented an extensive dermal melanocytosis, also known as Mongolian spot, involving the trunk, back, buttocks, upper and lower limbs, and abdominal erythematous patches, suggesting a capillary vascular malformation, namely port-wine stains (Figs. 1 and 2). The remaining physical examination was normal, and the newborn was asymptomatic. Ophthalmologic and neurologic evaluations as well as complementary diagnostic exams, including cranioencephalic magnetic resonance were normal. Systemic involvement was not detected and the lesions had not changed significantly over time. This congenital disorder was compatible with phacomatosis pigmentovascularis of cesioflammea type.

Phacomatosis pigmentovascularis is a rare congenital condition clinically defined by the presence of cutaneous vascular malformation and pigmented lesions.[1] It is a mosaic developmental abnormality of the vasomotor nerves and melanocytes (derived from the neural crest), caused by sporadic mutations in *GNA11* and *GNAQ* genes, which encode Gα subunits of heterotrimeric G proteins. These genetic mutations were detected in the affected tissues, but undetectable in the blood, suggesting that it was a post-zygotic mosaic disorder.[2]

The most recent classification divides phacomatosis pigmentovascularis into four groups, namely phacomatosis cesioflammea, spilorosea, cesiomarmorata, and unclassifiable phacomatosis pigmentovascularis. Phacomatosis cesioflammea is characterized by the presence of aberrant dermal melanocytosis and port-wine stains, with or without nevus anemicus.[1,3] Its possible association with extracutaneous

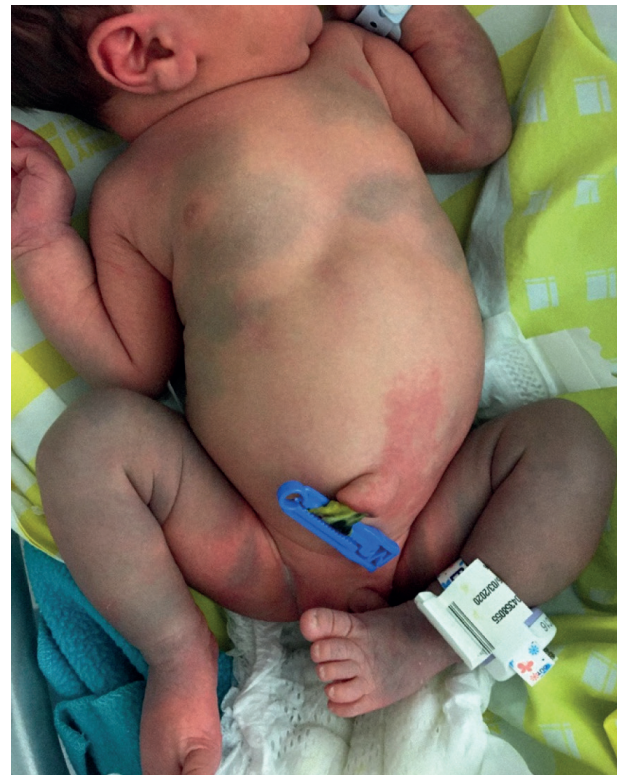

Figure 1. Abdominal port-wine stains or nevus flammeus.

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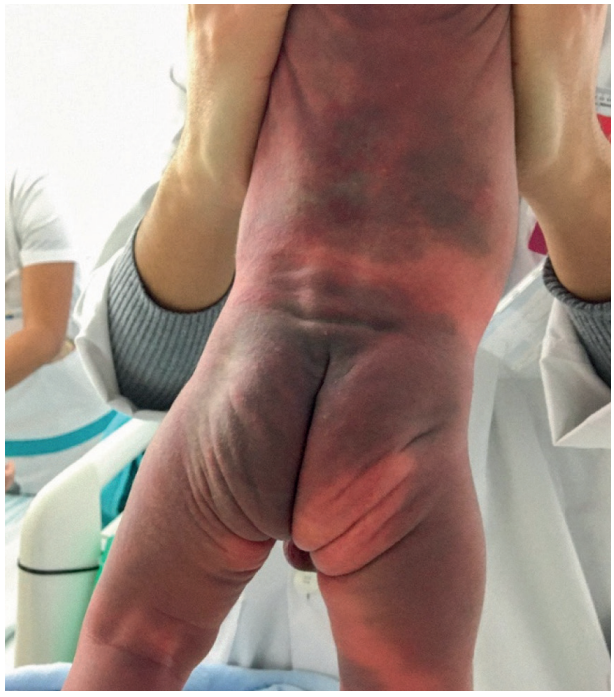

Figure 2. Extensive dermal melanocytosis.

manifestations, such as ocular, neurologic, and vascular manifestations, predicts its severity.[1,4,5] Therefore, an initial dermatologic, neurologic and ophthalmologic evaluation were suggested.[5] In the absence of systemic manifestations, phacomatosis pigmentovascularis is normally a benign condition and does not require any treatment. Nevertheless, the skin lesions might have a negative aesthetic impact and can be distressing for the children and their families. In those cases, laser treatment may be considered. Currently, the pulsed dye laser is the most accepted laser for port-wine stains.

Keywords: Newborn; Mongolian Spot/genetics; Neurocutaneous Syndromes/diagnosis; Port-Wine Stain/genetics; Skin Diseases, Genetic

Author Contribuitions

BA and IV participated in the study conception or design. BA and IV participated in acquisition of data. BA, IV and HC participated in the analysis or interpretation of data. BA, IV and HC participated in the drafting of the manuscript. BA, IV, MV and HC participated in the critical revision of the manuscript. All authors approved the final manuscript and are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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The authors declare that they have followed the protocols of their work centre on the publication of patient data.

Consent for publication

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