

Phacomatosis Pigmentovascularis: Case Report

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Abstract

Phacomatosis pigmentovascularis is defined as the association of a vascular malformation with an extensive pigment nevus. It is currently classified by Happle into three groups: cesioflamea, spilorosea and cesiomarmorata. We report a case of a female newborn presenting with typical manifestations of cesioflamea phacomatosis pigmentovascularis, according to the classification above. The rarity of this genodermatosis, the clinical exuberance of the skin lesions and the need for recognition of this syndrome and its repercussions motivated this case report.

Keywords: *Vascular Malformations; Pigmented Nevus; Mongolian Spot; Vascular Diseases*

Introduction

Pigmentovascular phacomatosis (PVF) is a rare congenital cutaneous syndrome. It is characterized mainly by the presence of capillary malformation and extensive pigmentary nevus and may or may not present with extracutaneous manifestations [1,2]. Systemic changes often involve abnormalities of the central nervous system, ocular and skeletal.

Case Report

A 5-day-old female infant, born full-term vaginally without complications, was evaluated by the Dermatology service for presenting, from birth, wine macules associated with diffuse blue-gray macules, affecting almost all of the dorsal body region. In some regions, there was overlapping of these cutaneous findings with hypochromic regions. In addition, on the face, he presented a pink spot that became more evident during crying. There was no cutaneous involvement in the anterior trunk, abdomen, genital region and ventral face of the upper and lower limbs.

During physical examination, no changes in neuropsychomotor development were observed and the body extremities were symmetrical, with no signs of atrophy or hypertrophy in the soft tissues.

Discussion

Pigmentovascular phacomatosis is a rare syndrome characterized by the concomitant presence of capillary malformation and pigmentary nevi, and is poorly described in the literature, with 245 cases described worldwide from 1947 to 2013, approximately [3].



Figure 1: Back of the patient evidencing the presence of extensive Mongolian spots, anemic nevus and port wine stains.



Figure 2: Salmon stain on the patient's face, which became more evident during crying.

The most recent ranking, created in 2005 by Happle, includes three main types:

- Phacomatosis cesioflammea: coexistence of Mongolian stain and stain in port wine, single or multiple;
- Phacomatosis spilorosea: coexistence of nevus spilus and salmon stain or, less often, the wine pattern of the stain in Port wine;
- Phacomatosis cesiomarmorata: coexistence of Mongolian spot and congenital telangiectasia marmorata cutis.

The coexistence of vascular and pigmented lesions that do not fit the above descriptions are considered as unclassifiable Type FPV. It is known that the majority (75%) patients have Cesioflammea phacomatosis [4,5].

The pathogenesis of PVF is still unclear, but it is believed to be an abnormality in the development of melanocytic cells and vasomotor neuronal cells derived from the neural crest [2]. The gene responsible for the syndrome has not yet been identified. Recently, somatic mutations of the GNAQ gene [guanine nucleotide binding protein (G protein), polypeptide q] have been identified in port wine stains and Sturge-Weber syndrome, indicating that there is a single underlying mechanism. There are still doubts whether GNAQ mutations or mutations of other genes in pluripotent progenitor cells are responsible for various Phenotypes of PvF [5].

Although the exact percentage is unclear, all forms of PPV may be associated with extracutaneous manifestations, such as central nervous system defects, ocular abnormalities, vascular malformations, limb asymmetry, unilateral lymphedema, and scoliosis. In addition to a complete dermatological examination, affected children should therefore also undergo pediatric and, if necessary, neurological, orthopedic and ophthalmological evaluation. This further helps to differentiate this condition from other vascular syndromes, including Sturge-Weber Syndrome (or encephalotrigeminal angiomatosis) and Klippel-Trenaunay syndrome [3].

In the case reported above, the wine lesions were compatible with port wine stains and were located in some regions of the back and buttocks. The grayish blue macules, located on the posterior trunk, back, buttocks and root of the limbs were compatible with extensive Mongolian spot. In some regions, there was overlapping of port wine spots and Mongolian spots with anemic nevus. In addition, the pink spot that became more evident during crying was characterized by salmon stain.

Based on cutaneous findings of Mongolian spots and capillary malformations, we then classify our patient as Cesioflammea phacomatosis, a clinical subtype found in most cases reported in the current literature.

Some systemic or cutaneous abnormalities may be associated with this type of phacomatosis, such as anemic nevus, alopecia, lower limb asymmetry, glaucoma, venous and lymphatic dysplasia, and syndromes such as Sturge-Weber and Klippel-Trenaunay.

In the present case, there was no parental consanguinity and reports of other similar cases in the family presenting vascular malformations.

Until the fifth month of life, the patient was in joint follow-up with pediatrics, and no extracutaneous alterations and signs and/or symptoms of other diseases or systemic involvement were found.

The treatment and prognosis of phacomatoses Pigmentovascular depend on the association with extracutaneous changes. As most cases occur sporadically without risk in subsequent pregnancies, prenatal screening is not necessary and this helps reassure parents during genetic counseling.

Since PVF without systemic involvement has a benign course and does not require treatment, in the case reported, we chose to perform expectant conduct, with periodic reassessment in order to detect early systemic signs of the disease.

Conclusion

In some cases, due to the impact on self-esteem, aesthetic procedures can be considered in order to improve the quality of life of patients, with no consensus until then on the exact moment for intervention. Some authors suggest that laser treatment should be done in childhood, ideally before school age, in order to reduce the extent of lesions [1-3].

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