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CASE REPORT

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.

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INTRODUCTION

Phacomatosis Pigmentovascularis (PPV) is a rare congenital cutaneous syndrome. It is mainly characterized by capillary malformation and extensive pigment nevus, which may or may not have extracutaneous manifestations[1,2]. Systemic changes often involve central nervous system, ocular and skeletal abnormalities.

CASE REPORT

A 5-day-old female infant, born at term, vaginally, without complications, was evaluated by the Dermatology Service for presenting, from birth, wine-colored macules associated with diffuse gray-blue macules, affecting almost all of the body's dorsal region. In some areas, these skin findings overlapped with hypochromic areas. In addition, on the face, it had 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 upper and lower limbs.

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

DISCUSSION

Phacomatosis pigmentovascularis is a rare syndrome characterized by the concomitant presence of capillary malformation and pigmentary nevi, which has been poorly described in the literature, with 245 cases described worldwide from 1947 to 2013, approximately[1].

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

Phacomatosis cesioflamea: single or multiple Mongolian and Port wine lesions coexistence;

Phacomatosis spilorosea: coexistence of nevus spilus and salmon stain or, less frequently, the wine stain pattern in port wine;

Phacomatosis cesiomarmorata: coexistence of Mongolian spot and congenital telangiectatic marble cutis.

The coexistence of vascular and pigmented lesions that do not fit the above descriptions are considered as unclassifiable PPV. Most patients (75%) are known to have Phacomatosis cesioflamea[3,4].

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

Although the exact percentage is not clear, all forms of PPV may be associated with extracutaneous manifestations, such as central nervous system defects, ocular anomalies, 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 ophthalmic 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-colored spots were compatible with port wine stains and were located in some regions of the back and buttocks. Blue-gray macules, located on the upper back, lower back, buttocks and root of the limbs, were compatible with extensive Mongolian spot. Portions of port wine and Mongolian patches with anemic nevus overlapped in some regions. In addition, the pink spot that became more evident during crying was characterized as the salmon spot.

Based on cutaneous findings of Mongolian spots and capillary malformations, we classified our patient as having Phacomatosis cesioflamea, a clinical subtype found in most cases reported in the current literature.

Some systemic or skin 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 case presented, there was no parental consanguinity and reports of other similar cases in the family presenting vascular malformations.

By the fifth month of life, the patient was under joint monitoring with pediatrics, and no extracutaneous changes and signs and/or symptoms of other diseases or systemic involvement were found.

Treatment and prognosis of phacomatosis pigmentovascularis depend on its 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.

Considering that PPV without systemic involvement has a benign course and does not require treatment, in this case, we chose expectant management, with periodic reassessment for an early detection of systemic signs of the disease.

In some cases, due to the impact on self-esteem, aesthetic procedures may be considered to improve the patients' quality of life, and there is no consensus as to the exact time for intervention. Some authors suggest that laser treatment should be done during childhood, ideally before school age, in order to reduce the extent of the lesions[1,2,5].

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