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Answer

A newborn with a rare association of various congenital skin disorders – Answer

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Answers

1. Port-wine stains (PWS) involving the infant's face, the upper left side of the chest, left shoulder, left arm, and most of the surface of her right leg and knee. Mongolian spots (MS) on the back intermingled with widespread cutis marmorata telangiectatica congenita (CMTC). PWS are congenital vascular malformations characterized by ectatic vessels in the upper dermis. Initially, they appear as pink macules, but then they tend to progressively darken, becoming reddish to purplish in nature [1]. MS are congenital single or multiple macular lesions typically affecting the sacrococcygeal or lumbar area. The color varies from blue to greenish and the size varies from few to more than 20 centimeters. Pigmentation gradually fades after the first year of life and it is rarely seen after the age of 6 years. MS usually resolve by early childhood and no treatment is generally needed [2]. CMTC is a rare congenital cutaneous vascular disorder. It usually presents at birth with persistent cutis marmorata, vascular telangiectasia, and occasionally ulcers. There is typically an improvement of the skin vascular pattern during the first year of life [3].
2. Phacomatosis cesioflammeo-marmorata.
3. Electroencephalogram (EEG), magnetic resonance imaging (MRI) of the brain and spinal cord, genetic analysis, and clinical follow-up.

Discussion

Phacomatosis pigmentovascularis (PPV) is a rare congenital disorder characterized by the coexistence of vascular and pigmentary nevus (MS, nevus of Ota, verrucous nevus, and nevus spilus) associated or not with extracutaneous manifestations [4].

PPV was first described by Ota in 1947, and over 250 cases of PPV have since been reported [4]. In 1985, Hasegawa and Yasuhara proposed a classification of PPV into four major types according to the combination of PWS and melanocytic lesion [5].

In 2005, Happle proposed a simpler classification and divided PPV into four types [6]:

1. phacomatosis cesioflammea (blue spots and nevus flammeus);
2. phacomatosis spilorosea (nevus spilus and light pink telangiectatic nevus);
3. phacomatosis cesiomarmorata (blue spots and cutis marmorata telangiectatica);
4. unclassifiable forms of phacomatosis.

The word cesioflammea is a Latin-derived compound noun: *caesius*, meaning blue-grayish, and *flammea*, which means fire or flame [7]. Therefore, phacomatosis cesioflammea is characterized by the coexistence of blue spots (dermal melanosis) and capillary malformations.

We report a female newborn with a combination of three skin lesions: MS, PWS, and CMTC. This association is an unclassifiable form of phacomatosis and only 7 similar cases have been published in the literature [4]. According to some authors, this association should be considered as a distinct entity of PPV with the specific name "phacomatosis cesioflammeo-marmorata" [8].

PPV pathogenesis is still unclear, but it is believed to be an abnormality in the development of melanocytic nevus cells and vasomotor neural cells derived from the neural crest [7]. The genetic phenomenon called twin spots or didymosis, a specific somatic mosaicism, has been suggested as a possible mechanism [4].

PPV is a sporadic disorder and it typically occurs for the first time in people with no family history of PPV [9]. PPV can be caused by a somatic mutation in the *GNA11* or *GNAQ* gene that is present only in the affected tissues of the body but not in other tissues or blood [10].

PPV may be associated with other systemic abnormalities in approximately 50% of all cases [11].

Other systemic or cutaneous abnormalities are anemic nevus, alopecia, lipohypoplasia, lower limb asymmetry, buphthalmos, dysplasia in veins and lymphatics, and syndromes such as Sturge-Weber and Klippel-Trenaunay [7].

The diagnosis of PPV is a gestalt diagnosis, based on the recognition of specific combined skin elements. The other tests (abdominal ultrasound, echocardiography, cranial ultrasound, MRI, EEG, and genetic analysis) are useful to exclude associated malformations, such as internal vascular anomalies, or syndromes.

In our patient, buphthalmos was present, but to date no other association has been demonstrated. MRI of the spine and brain has not yet been performed due to problems related to the patient's low age and the need for a specialized center. PPV without systematic complications is a benign condition and requires no treatment.

Laser treatment may improve a patient's quality of life, especially from an aesthetic point of view, by treating nevus flammeus and pigmentary nevus [7, 12].

Statement of Ethics

The patient's parents provided written informed consent. The study adhered to the tenets of the Declaration of Helsinki.

Declaration of interest

The Authors declare that there is no conflict of interest.

References

1. Huang Y, Yang J, Li Z, Zhang L, Sun L, Bi M, Wang L. Dermoscopic features of port-wine stains: A study of 264 cases. Australas J Dermatol. 2021;62(2):e201-6.
2. Gupta D, Thappa DM. Mongolian spots. Indian J Dermatol Venereol Leprol. 2013;79(4):469-78.
3. Shareef S, Alves JL, Horowitz D. Cutis Marmorata Telangiectatica Congenita. Treasure Island (FL): StatPearls Publishing, 2021.
4. Chehad AS. New case of phacomatosis cesio-flammeo-marmorata: the time is right to review the classification for phacomatosis pigmentovascularis. Int J Dermatol. 2019;58(12):e237-40.
5. Hasegawa Y, Yasuhara M. Phakomatosis pigmentovascularis type IVa. Arch Dermatol. 1985;121:651-5.
6. Happle R. Phacomatosis pigmentovascularis revisited and reclassified. Arch Dermatol. 2005;141:385-8.
7. Villarreal DJ, Leal F. Phacomatosis pigmentovascularis of cesioflammea type. An Bras Dermatol. 2016;91(5 Suppl 1):54-6.
8. Verma SB, Desai HK, Shah VN, Happle R. Phacomatosis cesioflammea with cutis marmorata-like lesions and unusual extracutaneous abnormalities: is it a distinct disorder? Indian J Dermatol. 2017;62:207-9.
9. Nanda A, Al-Abdulrazzaq HK, Habeeb YK, Zakkiriah M, Alghadhfan F, Al-Noun R, Al-Ajmi H. Phacomatosis pigmentovascularis: Report of four new cases. Indian J Dermatol Venereol Leprol. 2016;82(3):298-303.
10. Thomas AC, Zeng Z, Rivière JB, O'Shaughnessy R, Al-Olabi L, St-Onge J, Atherton DJ, Aubert H, Bagazgoitia L, Barbarot S, Bourrat E, Chiaverini C, Chong WK, Duffourd Y, Glover M, Groesser L, Hadj-Rabia S, Hamm H, Happle R, Mushtaq I, Lacour JP, Waelchli R, Wobser M, Vabres P, Patton EE, Kinsler VA. Mosaic Activating Mutations in GNA11 and GNAQ Are Associated with Phakomatosis Pigmentovascularis and Extensive Dermal Melanocytosis. J Invest Dermatol. 2016;136(4):770-8.
11. Adachi K, Togashi S, Sasaki K, Sekido M. Laser therapy treatment of phacomatosis pigmentovascularis type II: two case reports. J Med Case Rep. 2013;7:55.
12. Fernández-Guarino M, Boixeda P, de Las Heras E, Aboin S, García-Millán C, Olasolo PJ. Phakomatosis pigmentovascularis: Clinical findings in 15 patients and review of the literature. J Am Acad Dermatol. 2008;58(1):88-93.