PHACOMATOSIS PIGMENTOKERATOTICA. OBSERVATIONS ON A CASE

ALEXANDRU OANȚĂ*, SMARANDA ȚĂREAN**, VERONICA ILIESCU*, MARIA MAGDALENA CONSTANTIN***

Summary

The concept of twin spotting (didymosis) explains the combination of skin lesions in patients with Phacomatosis pigmentokeratotica.

We present the case of a 20-year-old patient with a verrucous congenital plaque located on the right retro-auricular region diagnosed as sebaceous nevus associated with Spillus nevus located contralaterally in the left temporal region.

Phacomatosis pigmentokeratotica represents the association of a organoid nevus with Spillus nevus differentiation. In addition, muscular hemiatrophy, neurological and ocular defects, deafness can be added to this association.

Key words: *phacomatosis pigmentokeratotica, spillus nevus.*

Received: 30.11.2018 *Accepted: 21.01.2019*

Introduction

The phacomatoses are congenital diseases simultaneously involving ocular, cutaneous and central nervous system affections with the appearance of malformations during development.

In 1996, Happle et al.[1] used the term of phacomatosis pigmentokeratotica (PPK) for a specific twin nevus syndrome in which spillus nevus (speckled lentiginous nevus - SLN) distributed as a checkerboard is associated with the organoid nevus (ON) with sebaceous differentiation, usually disposed linearly following the Blaschko lines, associated also with extracutaneous abnormalities: musculoskeletal, neurological and ocular[2].

Clinical case

We present the case of a 20-year-old male that was referred to our service for a congenital verrucous plaque located on the upper part of the right ear lobe (Fig.1). Biopsy from the plaque confirmed the diagnosis of sebaceous nevus. In addition, on the left side of the temporal region there was a tan lentiginous background patch on which there were more darkly pigmented macules distributed (Fig.2). The diagnosis of Spillus nevus was made.

Discussions

The concept of twin spotting (didymosis) explains the combination of cutaneous lesions in patients with phacomatosis pigmentokeratotica (PPK)[1,3].

In 1985, Jadahsson[4] introduced the term of „organoid naevus" (ON) for congenital lesions that have an excess or lack of skin components. In 1932, Robinson[5] defines sebaceous nevus of Jadahsson as a lesion localized on the face and scalp characterized by papillomatous epidermal hyperplasia and an increased number of hypertrophic sebaceous glands.

* Dermamed, Brasov.
** TopMed, Targu Mures.
*** Clinical Hospital Colentina, Dermatology 2, Bucharest.

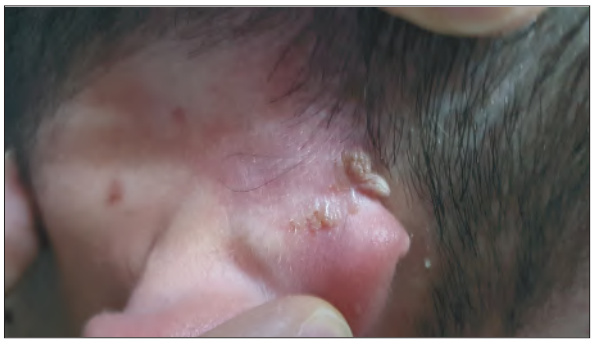

Fig. 1. Clinical appearance of sebaceous nect

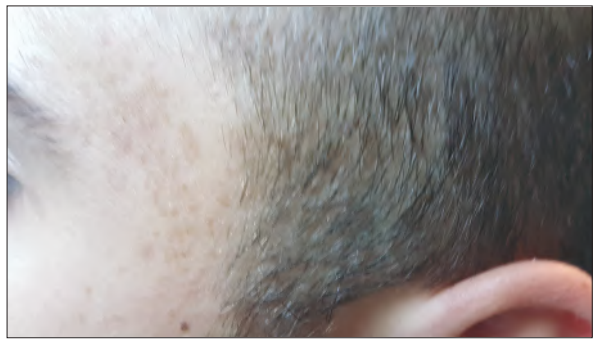

Fig. 2. Clinical appearance of Spillus nevus

In 1995, Tadini et al[6] presents the case of a 9-year-old girl with unilaterally located spillus nevus (SLN) associated with an epidermal verrucous nevus located contralaterally and a ichthyosis-like diffuse hyperkeratosis. The same authors, studying several cases, define PPK in 1998 as an association of ON with sebaceous differentiation located on the Blaschko lines with SLN arranged as a checkerboard[7]. The two lesions may be associated with muscular hemiatrophy accompanied by muscular weakness of different degrees, neurological defects (segmental dysaesthesia, hyperhidrosis, mild mental retardation, epileptic seizures), ocular defects (ptosis, strabismus), deafness.

Although a number of authors found a predominantly contralateral distribution of SLN to ON as in our case, it appears that this distribution is present in only 20% of the PPK reported cases[8]. SLN and ON seem to be located ipsilaterally in 33% of PPK cases, and SLN, ON or both are bilaterally distributed in 40%, 3% and 3%, respectively. Locations of SLN and especially of ON are more common on the head and neck. In PPK, on the SLN are found different types of nevi, such as common nevi (jonctional, dermal, compound), dysplastic nevus, blue nevus, spitz nevus or congenital nevus[2,7,9,10,11,12,13]. It was described the possibility of melanoma on SLN[7,13] and basal cell carcinoma on ON[9,13,14,15].

Solomon[16] introduced in 1968 the concept of epidermal nevus syndrome, unrecognized by the fact that the epidermal nevus was associated with abnormalities of a variety of organs. In this group there are distinct phenotypic syndromes such as sebaceous nevus syndrome (Schimmelpenning syndrome), PROTEUS syndrome, nevus comedonicus syndrome, Becker's syndrome.

Regarding the differential diagnosis of PPK, this is done with sebaceous neovascular syndrome (Schimmelpenning syndrome) that associates ON with sebaceous differentiation with ocular, cerebral and skeletal manifestations. PPK is readily differentiated from pigmented vascular facial surgery characterized by the simultaneous emergence of expanded telangiectasic nevus with expanded pigment nev (non-epidermal linear, dermal melanosis, nev spilus).

There is no causal therapy in PPK. Symptomatic improvements can be achieved through dermabrasion, surgical excision, laser therapy (Erbium YAG laser). Our case presents the association of SLN with ON without affecting other organs, recommending that the excision of the sebaceous nevus should be made.

In conclusion, patients with PPK should be carefully examinated for organ damage and correct assessment.

Bibliography

1. Happle R, Hoffmann R, Restano L, Caputo R, Tadini G. Phacomatosis pigmentokeratotica: a melanocytic-epidermal twin nevus syndrome. Am J Med Genet. 1996 Nov 11;65(4):363-5.
2. Sugarman GI, Reed WB. Two unusual neurocutaneous disorders with facial cutaneous signs.Arch Neurol 1969. Sep;21(3):242-7.
3. Wauschkuhn J, Rohde B. Systematized sebaceous, pigmented and epithelial nevi with neurologic symptoms. Neuroectodermal Feuerstein-Mims syndrome. Hautarzt.1971 Jan;22(1):10-3.
4. Jadassohn J. Bemerkungen zur Histologie der systematisierten Naevi und über "Talgdrüsen- Naevi". Arch Derm Syphilol 1895;33:355-95.
5. Robinson SS. Nevus sebaceous (Jadassohn): Report of four cases. Arch Dermatol Syph.1932;26:633.
6. Tadini G, Ermacora E. Carminati G, Gelmetti C, Cambiaghi S, Brusasco A, Caputo R, Happle R. Unilateral speckled-lentiginous nevus, contralateral verrucous epidermal naevus, and diffuse ichthyosis-like hyperkeratosis: An unusual example of twin spotting? Eur j Dermatol 1995;5:659-663.
7. Tadini G, Restano L, Gonzáles-Pérez R, Gonzáles-Enseñat A, Vincente-Villa MA, Cambiaghi S, et al. Phacomatosis pigmentokeratotica: report of new cases and further delineation of the syndrome. Arch Dermatol. 1998 Mar; 134(3):333-7.
8. Gruson LM, Orlow SJ, Schaffer JV. Phacomatosis pigmentokeratotica associated with hemihypertrophy and a rhabdomyosarcoma of the abdominal wall. J Am Acad Dermatol. 2006 Aug;55(2 Suppl):S16-20.
9. Goldblum JR, Headington JT. Hypophosphatemic vitamin D-resistant rickets and multiple spindle and epithelioid nevi associated with linear nevus sebaceus syndrome. J Am Acad Dermatol. 1993 Jul;29(1):109-11.
10. Hill VA, Felix RH, Mortimer PS, Harper JI. Phacomatosis pigmentokeratotica. J R Soc Med. 2003 Jan;96(1):30-1.
11. Okada E, Tamura A, Ishikawa O. Phacomatosis pigmentokeratotica complicated with juvenile onset hypertension. Acta Derm Venereol. 2004;84(5):397-8.
12. Vidaurri-de la Cruz H, Tamayo-Sánchez L, Durán-McKinster C, de la Luz Orozco-Covarrubias M, Ruiz-Maldonado R. Epidermal nevus syndromes: clinical findings in 35 patients. Pediatr Dermatol. 2004 Jul-Aug;21(4):432-9.
13. Martinez-Menchón T, Mahiques Santos L, Vilata Corell JJ, Febrer Bosch I, Fortea Baixauli JM. Phacomatosis pigmentokeratotica: a 20-year follow-up with malignant degeneration of both nevus components. Pediatr Dermatol. 2005 Jan-Feb;22(1):44-7.
14. Misago N, Narisawa Y, Nishi T, Kohda H. Association of nevus sebaceus with an unusual type of „combined nevus". J Cutan Pathol. 1994 Feb;21(1):76-81.
15. Langenbach N, Hohenleutner U, Landthaler M. Phacomatosis pigmentokeratotica: speckled lentiginous nevus in association with nevus sebaceus. Dermatology. 1998;197(4):377-80.
16. Solomon LM, Fretzin DF, Dewald RL. The epidermal nevus syndrome. Arch Dermatol. 1968;97:273.

Conflict of interest
NONE DECLARED

Correspondance address: Alexandru Oanță
Dermamed Brasov
E-mail: oanta_alexandru@yahoo.com