

CLINICAL REPORT

Phacomatosis Pigmentokeratotica (Happle) in a 23-Year-Old Man

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Phacomatosis pigmentokeratotica is a rare but highly characteristic disease defined by the occurrence of an organoid naevus with sebaceous differentiation, a speckled-lentiginous naevus and other associated anomalies. It is probably caused by the twin-spot phenomenon. We report on a 23-year-old male electrician with 10 irregularly shaped, sharply demarcated, brownish-yellow papillomatous plaques following Blaschko's lines, as well as 6 large, sharply demarcated, round to oval, slightly greyish macules with pewit-egg-like dots, involving both buttocks, the right thigh, the right knee, the right pectoral region and the upper back. A moderate hyperhidrosis of the palms, soles and axillae was noted. All routine blood tests and laboratory findings, including chest X-ray, ECG, abdominal ultrasound, ocular and neural examination were unrevealing. Phacomatosis pigmentokeratotica may be associated with dysaesthesia, segmental hyperhidrosis, mild mental retardation, epileptic seizures, deafness, ptosis, strabismus or muscular weakness. In our patient, only slight hyperhidrosis was present, whereas all other associated anomalies could be excluded. **Key words:** epidermal naevus; lines of Blaschko; naevus spilus; twin spotting.

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The association of a speckled lentiginous naevus, an organoid naevus with sebaceous differentiation and skeletal and neurologic anomalies constitutes a specific syndrome that has been called phacomatosis pigmentokeratotica (PPK) (1). It has been hypothesized that the co-occurrence of the two different naevi reflects a twin-spot phenomenon (2, 3). The concept of twin spotting is a postzygotic crossing-over in a doubly heterozygous embryo at an early stage of embryogenesis, resulting in 2 different mutated stem cells generating 2 types of naevi in a mosaic pattern (see ref 6 and 7 below for a detailed explanation). We describe a patient affected by this rare syndrome.

CASE REPORT

A 23-year-old male electrician had a history of congenital skin lesions that had grown in size with the patient. He complained of no other symptoms apart from the cosmetic impairment. About 10 irregularly formed but sharply demarcated, yellowish-brown, confluent papillomatous plaques were seen on the scalp, face, neck and sternal region; all followed Blaschko's lines (Fig. 1a–d). In addition, about 6 large, sharply demarcated, round to oval, slightly greyish macules with pewit-egg-like dark-brown dots were seen on both buttocks, the left thigh, right knee, right pectoral region and upper back. Routine blood tests and other laboratory findings, including chest X-ray, ECG, abdominal ultrasound, ocular and neurological examination, gave normal results. Orthopedic examination showed slight scoliosis, but was otherwise unrevealing. A moderate hyperhidrosis of the palms, the soles and the axillae was present. Histopathological examination of two biopsy specimens obtained from the organoid naevus, involving the parietal and occipital region of the scalp, showed pigmented trichoblastoma. A biopsy obtained from a dark dot within the speckled-lentiginous naevus from the upper back displayed features of a dysplastic melanocytic naevus cell of the junctional type. Part of the organoid naevus on the right mandibula was test-treated with an Erbium-YAG laser. The short-term results after 6 months are fairly satisfactory, so we decided to continue the treatment.

DISCUSSION

In 1895, Jadassohn introduced the term *organ naevus* for circumscribed congenital lesions consisting of excess or deficiencies of normal skin constituents (4). In 1932, Robinson defined the term *sebaceous naevus of Jadassohn* as localized lesions of the face and scalp characterized by papillomatous epidermal hyperplasia and an excessive number of enlarged sebaceous glands (5). In 1995, Tadini et al. reported an unusual type of a unilateral speckled lentiginous naevus with a contralateral verrucous epidermal naevus and a diffuse ichthyosis-like hyperkeratosis in a 9-year-old girl (6). In 1998 the same authors reported additional cases of PPK, with findings that allowed them to better delineate this syndrome. They gave the following definition for PPK: multiple organoid naevi with sebaceous differentiation, arranged



Fig. 1. Phacomatosis pigmentokeratotica. A systematized naevus sebaceus (a, b) involving head, neck and trunk is seen in a pattern following Blaschko's lines. In addition, a systematized speckled lentiginous naevus is present (c, d), arranged in a checkerboard distribution on the chest.

according to Blaschko lines, along with a speckled-lentiginous naevus arranged in a checkerboard pattern (7). In addition, hemiatrophy with muscular weakness

of varying degrees and other neurologic defects may be present, e.g., segmental dysaesthesia, hyperhidrosis, mild mental retardation, epileptic seizures, deafness, ptosis,

strabismus and muscular weakness of varying degrees (7–10). In our patient, only slight hyperhidrosis was found, whereas all other associated diseases could be excluded.

For differential diagnosis, epidermal naevus syndromes such as Schimmelpenning syndrome may be considered. Schimmelpenning syndrome is characterized by an organoid naevus showing sebaceous differentiation in association with ocular, cerebral and skeletal defects. By contrast, PPK consists of an organoid naevus that usually shows a sebaceous differentiation and is arranged in a systematized linear pattern, in association with a speckled-lentiginous naevus distributed in a checkerboard pattern, as well as hemiatrophy and neurologic defects. PPK and phacomatosis pigmentovascularis can be distinguished readily, because the latter syndrome is characterized by the simultaneous occurrence of a widespread telangiectatic naevus and an extensive pigmentary naevus. In order to recognize a neurological involvement that is often present in PPK, a neurological examination is necessary. Patients must be monitored for the occurrence of trichoblastoma in the organoid naevi on a regular basis.

There is no causal therapy of PPK. Potential methods for a symptomatic improvement of the lesions are dermabrasion, excision or laser therapy. In our patient, symptomatic Erbium-YAG laser therapy was chosen and yielded satisfactory results.

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