Keratinocytic Epidermal Nevus with Nevus Spilus and Underlying Hemi hypertrophy

Hemihypertrofinin Eşlik Ettiği Nevus Spilus ve Keratinositik Epidermal Nevüs Birlikteliği

ABSTRACT Epidermal nevi are congenital hamartomas originated from ectodermal cells of embryo. Nevus spilus is an epidermal nevus consisting of a light tan patch with numerous dark brown macules and papules within it. The term ‘epidermal nevus syndrome’ is used to describe the association of epidermal hamartomas and extra-cutaneous abnormalities. Here we present a rare case of a 5-year-old girl with a keratinocytic epidermal nevus and nevus spilus accompanying hemi-hypertrophy which is not consistent with any known epidermal nevus syndromes.

Keywords: Epidermal nevus syndrome; hemi-hypertrophy; keratinocytic epidermal nevus; nevus spilus


Anahat Karime: Epidermal nevus sendromu; hemi-hipertrofi; keratinositik epidermal nevüs; nevus spilus

CASE REPORT

A 5-year-old girl presented with a brown verrucous plaque lesion on her neck which had been present since birth. The parents of the patient also had the complaint of darkening of the color in the patient’s right leg. Her mental and motor functions were within the normal limits except the his-
tory of late onset of gait. The patient’s parents reported similar lesions in two family relatives of the patient.

Dermatologic examination revealed, yellow-brown, verrucous plaque in 10cm length and 0.3-2cm width on the neck (Figure 1). A hyperpigmented patch lesion on the right side of the body that was limited by the midline zone and extended to all over the right leg was present (Figure 2). Numerous black and brown macules and papules of varying sizes were present within the affected area. Hemi-hypertrophy was also noted on the right limb.

The family refused to let us perform biopsies for histopathological examination. Based on the clinical appearance the verrucous lesion on the neck was diagnosed as ‘keratinocytic epidermal nevus’ and the one along the lower extremity as ‘nevus spilus’.

The radiographic examination revealed lengthening and increase in thickness of the bony structures of the hypertrophic side compared with the left side. Physical and neurologic examination showed no neurological, skeletal, or other extracutaneous abnormalities. The common blood count, biochemical tests, calcium and phosphor levels in 24 hour urine sample, abdominal ultrasonography, cranial magnetic resonance imaging, electroencephalography were within the normal limits. The patient showed no features related with any ENSs.

Written consent was obtained from the parents of the patient for publication of this case report and any accompanying images.

DISCUSSION

Epidermal nevi (EN) are hamartomas of keratinocytic or epidermal appendages. Epidermal nevi are congenital hamartomatous lesions derived from epidermal components originating in pluripo-
tential cell mutations during early embryonic stage. ENSs are defined as the presence of an EN with associated involvement of other organ systems. In recent years, data on different ENSs has expanded, and in some of these phenotypes the underlying molecular defects, clinical features and genetic patterns has been elucidated. All ENSs are reported to reflect mosaicism. Although most of them, exclusively occur sporadically, there are also hereditary traits. Well known ENSs include Schimelpenning syndrome, phacomatosis pigmentokeratotica (PPK), nevus comedonicus syndrome, angora hair nevus syndrome, Becker nevus syndrome, Proteus syndrome and Congenital hemidysplasia with ichthyosiform nevus and limb defects (CHILD) syndrome.

The patient, presented here, had keratinocytic nevus on the neck and nevus spilus on the leg. Nevus spilus (speckled lentiginous nevus) is a variant of congenital melanocytic nevus consisting of a light tan patch with numerous dark brown macules and papules. Recently, macular and papular types of nevus spilus has been described as different histopathological entities and each of them indicates different phacomatosis types. It is considered that macular type of nevus spilus is associated with phacomatosis pigmentovascularis while papular type of nevus spilus accompanies PPK.

PPK, one of the ENSs, is characterized by a coexistence of nevus sebaceous that is usually rather large and a papular nevus spilus (speckled lentiginous nevus of the papular type). While epidermal nevus presents with Blaschko pattern, the melanocytic component usually occurs in checkerboard pattern. Central nervous system defects, including mental deficiency, seizures, hemiparesis, and hyperhydrosis, disesthesia, sensory or motor neuropathy are reported to be characteristic neurologic features of the associated papular nevus spilus syndrome. Additionally, PPK has been reported to be associated with bony asymmetries including hypoplasia of the pelvis, kyphoscoliosis and hemi-atrophy. Hemiatrophy of tissue underlying the nevi with muscular weakness appears to be a common finding.

The patient had both macular and papular types of nevus spilus and hemi-hypertrophy, suggesting the diagnosis of PPK. However, she didn’t have nevus sebaceous, which is the component of the PPK. Instead, the plaque lesion on the patient’s neck was consistent with keratinocytic nevus in clinical appearance.

The cooccurrence of keratinocytic nevus and nevus spilus is rarely reported. Both the keratinocytic nevus and the nevus spilus the patient had, presented within the blaschkoid pattern, which suggests a common pathogenesis in the development of lesions. Although it is still unclarified, the lesions following Blaschko lines are believed to develop from a genetic mosaicism. The patient showed no extracutaneous anomalies, other than hemi-hypertrophy of the right limb, on that also nevus spilus is present.

Keratinocytic epidermal nevus, nevus spilus and hemihypertrophy cooccurrence is not consistent with any of the known ENSs. To the best of our knowledge there is no current data in the English literature reporting the association of keratinocytic epidermal nevus, nevus spilus and hemihypertrophy. It may be hypothesized that during the follow-up new symptoms in the skin or in other systems may develop that may lead the diagnosis of PPK.

Unfortunately as the parents of the patient did not give consent, histopathologic examination could not be performed. Since nevus spilus and epidermal nevus can be diagnosed based on the clinical appearance, we believe that the lack of histopathologic examination may be assumed as a minor limitation. The case is unique with cooccurrence of keratinocytic epidermal nevus, nevus spilus and hemihypertrophy which might eventually turn into diagnosis of a epidermal nevus syndrome during the follow-up.

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