

Systematized epidermal nevus: A case report

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ABSTRACT:

Systematized epidermal nevus (SEN) is an extensively distributed variant of keratinocytic epidermal nevi. The clinical features include bilateral well-defined skin-colored to hyperpigmented verrucous or velvety papules that coalesce to form plaques in a Blaschkoid distribution. We report a case of SEN in an eight-year-old Thai boy. He presented with multiple brownish hyperkeratotic papules and plaques on his face, neck, chest, and back arranged along the lines of Blaschko with no evidence of systemic involvement. The diagnosis was supported by clinical manifestations and histopathological features. The patient received multiple sessions of carbon dioxide (CO₂) laser. Long-term follow-up is necessary throughout the patient's lifetime, as neurological manifestations may present later in adulthood.

Key words: Epidermal nevus, epidermal nevus syndrome, systematized epidermal nevus

Case Report

An eight-year-old Thai boy presented with multiple brownish hyperkeratotic papules and plaques on his face, neck, upper chest, buttocks and back. The lesions appeared when he was 6 months old and developed more hyperpigmentation and thickening with age. Developmental milestones were normal. Family history was non-contributory.

Physical examination revealed that there were multiple bilateral linear streaks of brownish hyperkeratotic papules and plaques along the lines of Blaschko on the neck, upper chest, and back (Figure 1). Hair, teeth, and nails were normal. The eye examination was normal. No systemic abnormalities were detected on clinical examination. The genetic testing was not done due to financial issues.



Figure 1 Multiple bilateral linear streaks of brownish hyperkeratotic papules and plaques along the lines of Blaschko on (A) neck, (B) right upper chest and (C) back

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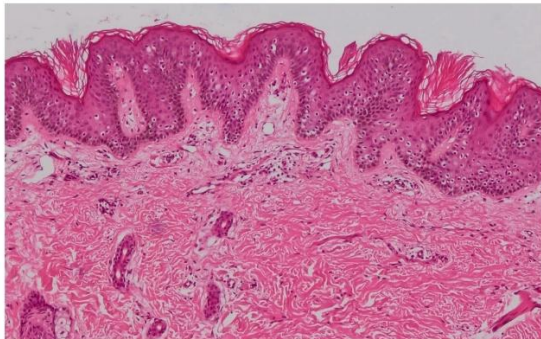


Figure 2 Hyperplasia of the epidermis with hypermelanization and hyperkeratosis with subtle inflammation in dermis (H&E, X100)

Differential diagnosis is systematized epidermal nevus, inflammatory linear verrucous

epidermal nevus, linear psoriasis, and lichen striatus.

A histological examination of his back revealed hyperplasia of the epidermis with hypermelanization and hyperkeratosis with subtle inflammation in the dermis (Figure 2). A diagnosis of systematized epidermal nevus was made by clinical and histopathological examination.

The patient was treated with ablative carbon dioxide (CO₂) laser therapy for four sessions on the neck within a two-year follow-up period. The treated lesions tended to improve over multiple sessions of therapy. However other lesions became spreading and worsening with time.

Table 1 Classification of epidermal nevus syndrome

Well-defined phenotypes	Less clearly defined phenotypes
Organoid nevi • Schimmelpenning syndrome • Phacomatosis pigmentokeratotic • Nevus comedonicus syndrome • Angora hair nevus syndrome • Becker nevus syndrome Keratinocytic nevi • CHILD syndrome (congenital hemiplasia with ichthyosiform nevus and limb defects) • Type 2 segmental Cowden disease • <i>FGFR3</i> epidermal nevus syndrome (Garcia-Hafner-Happle syndrome) • Proteus syndrome	• Nevus trichilemmocysticus syndrome • Didymosis aplasticosebacea • SCALP syndrome (a combination of sebaceous nevus, CNS abnormalities, aplasia cutis, limbal dermoid, and pigmented nevi) • Gobello syndrome • Bäfverstedt syndrome • NEVADA syndrome (epidermal nevus with vascular malformations) • CLOVE syndrome (congenital lipomatous overgrowth with vascular anomalies and epidermal nevus)

Adapted from: Happle R. The group of epidermal nevus syndromes Part I&II^{6,7}

Discussion

Epidermal nevi originate from benign overgrowth of embryonic ectoderm and present at birth or within the early years of life^{1,2}. The incidence of epidermal nevi ranges from 1 to 3 per 1000 live births, affecting males and females equally³. Epidermal nevi usually occur sporadically. The appearance of these lesions is due to postzygotic mutations of *FGFR3*,

PIK3CA, or *HRAS* genes in a subset of the skin cell lineage, while the rest of the cells are normal, leading to somatic mosaicism. This results in abnormal epidermal growth limited to certain areas⁴.

Epidermal nevi can present in isolation or in spectrums and are associated with a wide variety of disorders. Systematized epidermal nevus refers to extensive epidermal nevus

lesions on the skin as bilateral linear and whorled tan patches or plaques distributed along the lines of Blaschko without any other systemic abnormalities. Nevus unius lateris is a rare variant of systematized epidermal nevus distinguished by its extensive unilateral distribution⁵. In the early stage, the lesions appear as patches. Over time, they become thickened or verrucous plaques and persist lifelong. In contrast, epidermal nevus syndrome (ENS) refers to the combination of an epidermal nevus (often extensive) with associated systemic abnormalities. Thorough serial examinations should be performed to observe for neurologic deficits (seizures, cognitive impairment), skeletal anomalies (bone deformities, asymmetry, limb length discrepancy), ocular abnormalities (colobomas, vision loss), and occasionally visceral anomalies (heart defects, kidney problems, etc.)^{2, 6}.

ENS represents a group of distinct disorders that can be classified by the type of associated epidermal nevi and by the criterion of presence or absence of heritability. Well-defined and less well-defined phenotypes for these disorders are categorized in Table 1.

Histology of epidermal nevus is characterized by acanthosis, orthohyperkeratosis, papillomatosis, and an expanded papillary dermis which is sharply demarcated from the surrounding normal skin¹.

Management of small isolated epidermal nevi in patients with a normal physical examination usually does not require further work-up. In contrast to extensive epidermal nevi, the screening laboratory investigation should be performed if any systemic abnormalities are present. A multidisciplinary approach may need to be adopted depending on the type and extent of other organ involvement¹.

In an isolated systematized epidermal nevus, various treatment options were applied to the patients for cosmetic concerns, such as topical keratolytics, topical calcipotriol, topical

tretinoin, podophyllin, 5-fluorouracil, cryotherapy, CO₂ laser ablation, and surgical removal^{1,4,5}. Recurrence is common with topical treatment. Both destructive therapies and surgical excision carry a risk of scarring. While CO₂ laser ablation is less invasive than surgery, improper depth of the laser can lead to either recurrence or hypertrophic scarring⁸. The choices of treatment may be considering the age of the patient, and the extent of lesions. Large extensive lesions may be difficult to remove completely. Oral retinoids are one of the treatment options; however, the response varies.⁹

There are reports of secondary skin cancers arising within epidermal nevi and neurological abnormalities that present in late adulthood^{2, 10}. We recommend serial observations for any changes in the lesions and thorough examinations are warranted. In clinical practice, the patient who presents with extensive lesions of epidermal nevus should be screened for systemic organ abnormalities. Early detection of epidermal nevus syndrome yields timely appropriate management^{6, 7}.

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