

Progressive symmetrical Erythrokeratoderma: A case report and literature review.

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ABSTRACT:

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Progressive symmetrical erythrokeratoderma (PSEK) is a rare disorder of cornification characterized by epidermal hyperproliferation with predominantly autosomal dominant inheritance, and sometimes autosomal recessive transmission has been also observed. The molecular basis of PSEK in the vast majority of patients has not yet been established. Patients with PSEK usually respond to oral retinoid therapy. In this report, we describe the patient with PSEK. A 20-year old female with multiple hyperpigmented patches on neck, back and lower legs since birth. Histopathological finding is compatible with progressive symmetrical erythrokeratoderma.

Key words: Progressive symmetrical erythrokeratoderma (PSEK), loricrin, connexin

บทคัดย่อ :

ภาสุร์ เปี่ยมพงศ์สานต์ โกวิท คัมภีรภาพ รายงานผู้ป่วยและการทบทวนวรรณกรรมโรค **PROGRESSIVE SYMMETRICAL ERYTHROKERATODERMA** วารสารโรคผิวหนัง 2553; 26: 214-218.

สถาบันโรคผิวหนัง กรมการแพทย์ กระทรวงสาธารณสุข

Progressive symmetrical erythrokeratoderma เป็นโรคที่เกิดจากการเพิ่มจำนวนอย่างผิดปกติของเซลล์ผิวหนังชั้นหนังกำพร้า การถ่ายทอดทางพันธุกรรมส่วนใหญ่เป็นแบบยีนเด่น แต่ก็อาจพบการถ่ายทอดแบบยีนด้อยได้ด้วย ลักษณะพื้นฐานทางโมเลกุลของโรคนี้ในคนไข้ส่วนมากยังไม่สามารถพิสูจน์แน่ชัด การรักษาด้วยยาเรตินอยด์ชนิดรับประทานได้ผลดี รายงานฉบับนี้เป็นการนำเสนอผู้ป่วยหญิงอายุ 20 ปี มาตรวจด้วยผื่นสีเข้มที่บริเวณคอ หลัง และขาทั้งสองข้างแต่กำเนิดผลตรวจทางจุลพยาธิเข้าได้กับโรค Progressive symmetrical erythrokeratoderma

คำสำคัญ: การเพิ่มจำนวนอย่างผิดปกติของเซลล์ผิวหนังชั้นหนังกำพร้า

Case report

A 20-year-old Thai female patient from Ayutthaya presented with multiple hyperpigmented patches on neck, back and lower legs since birth. Since birth she has had non-pruritic erythematous patches on hands, legs, and back. Later they became thicker and expanding. No other underlying disease was noted. Her child had similar skin lesions, which improved by the age of two.

An examination revealed multiple well-defined, discrete, symmetrical, fine scaly, erythematous and hyperpigmented patches on neck, back, arms, and lower legs (Figures 1). No other abnormality detected.

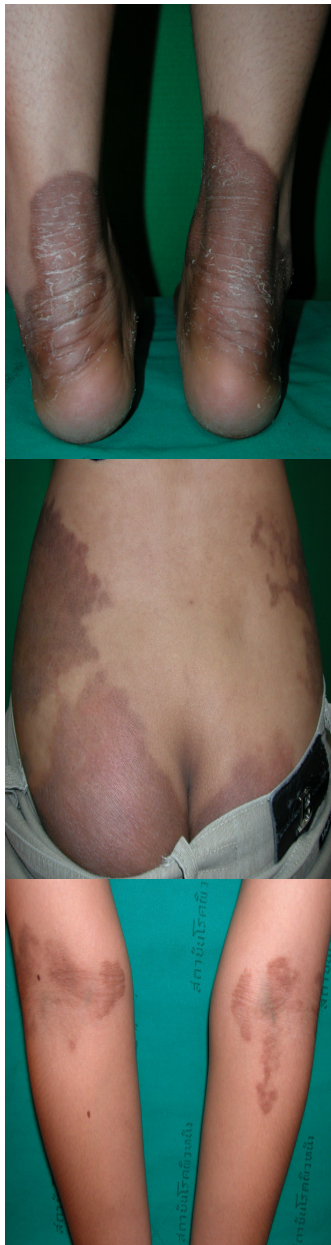
Histopathological finding showed marked compact hyperkeratosis, epidermal hyperplasia in association with superficial perivascular inflammatory cell infiltration of lymphocytes (Figures 2). Provisional

diagnosis was progressive symmetrical erythrokeratoderma.

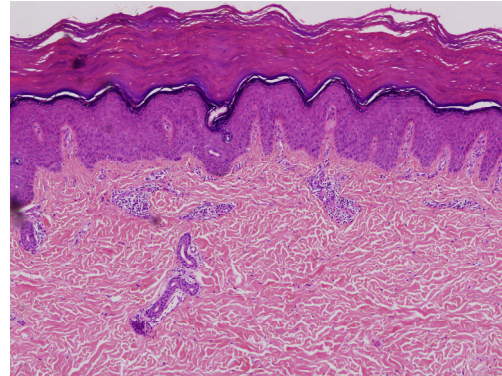
Discussion

Progressive symmetrical erythrokeratoderma (PSEK) is a rare genodermatosis which clinically heterogenous but usually characterized by fixed and slowly progressive erythematous and hyperkeratotic plaques distributed symmetrically over the trunk, particularly on the limbs and buttock.¹ The face, palms and soles may also be involved. Onset usually begins during infancy or early childhood, progresses and increase in number and size overtime, but tend to stabilize after puberty. It was first described by Darier in 1911² and defined as PSEK by Gottron later.³ There is no sexual predilection. Patients are otherwise mentally and physically unaffected. PSEK is inherited as an autosomal dominant trait with incomplete

penetrance and variable expressivity^{4,5}, although autosomal recessive transmission has been also observed.⁶



Figures 1



Figures 2

The molecular basis of PSEK in the vast majority of patients has not yet been established.^{1,7-9} Ishida-Yamamoto et al.⁹ found that affected members of a Japanese family with PSEK and mutilating palmoplantar keratoderma with pseudoainhum of the Camisa type had a mutation in the loricrin gene (LOR), which maps to 1q21.

Loricrin is the major structural component of the cornified cell envelope that is formed beneath the plasma membrane of stratified squamous epithelial cells during terminal differentiation. Loricrin mutations have also been found in Vohwinkel syndrome¹⁰ and in a congenital ichthyosiform erythroderma presenting as a collodion baby and later developing palmoplantar keratoderma and pseudoainhum.¹¹

Cui et al.¹² recently showed a novel locus for PSEK on chromosome 21q11.2-21q21.2 in a Chinese family, but no gene has been identified to date. Akman et al.¹³ report the first Turkish patient. Their molecular studies of the loricrin (LOR), connexin 31 (GJB3) and

connexin 30.3 (GJB4) genes did not identify a disease-causing mutation.

Histologic finding, PSEK shows acanthosis of the epidermis with basket-weave pattern and often patchy parakeratotic hyperkeratosis. The granular layer is prominent and sometimes shows intracellular vacuolization. Follicular plugging is not common. Some acanthosis and papillomatosis and occasional dyskeratotic epidermal cells can occur.¹⁴ Dilated capillaries and sparse lymphocytic perivascular infiltrates are found in the papillary dermis.¹³ PSEK should be a clinical diagnosis, since the microscopic features are nonspecific. Therefore, our patient was diagnosed with PSEK based on clinical and histological findings.

The clinical differential diagnosis of PSEK include erythrokeratoderma with ataxia, erythrokeratoderma variabilis(EKV), psoriasis and pityriasis rubra pilaris(PRP). Erythrokeratoderma with ataxia, in contrast to PSEK, there are only a few, slightly erythematous, hyperkeratotic plaques on ankles, knees and elbows that diminish or disappear during adulthood, while progressive spinocerebellar ataxia develops later in life. The occurrence of transient, variable erythema allows EKV different from PSEK. The presence of perifollicular papules with an erythematous base, a coalescence of orange-red plaques, but with obvious islands of sparing and the orange-red waxy keratoderma of the palm and sole can help to distinguish PRP. Psoriasis can usually be excluded by its silvery scales,

nail findings and typical abnormalities seen on light microscopic skin examination.¹³

Symptomatic treatment with topical keratolytics or emollients is usually not successful. Several PSEK patients respond very well to systemic treatment with retinoids. While others reported that PUVA therapy was equally effective.^{15,16}

PSEK is a rare skin disease. We diagnosed our patient as having PSEK by her typical clinical features and histologic findings. The patient did not receive oral retinoid and partially respond to treatment with topical keratolytics and topical emollients.

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