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
Progressive symmetrical erythrokeratoderma is a rare genodermatosis which clinically heterogeneous but usually characterized by fixed and slowly progressive erythematous and hyperkeratotic plaques distributed symmetrically over the trunk, particularly on the limbs and buttock. 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 1911 and defined as PSEK by Gottron later. 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**

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 syndrome\(^10\) 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. A 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. Dilated capillaries and sparse lymphocytic perivascular infiltrates are found in the papillary dermis. 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 erythrokeratodermia with ataxia, erythrokeratodermia variabilis (EKV), psoriasis and pityriasis rubra pilaris (PRP). Erythrokeratodermia 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.

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.

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.

References


