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Erythrokeratodermia Progressiva Symmetrica: A Case Report from the Dermatology Hospital of Bamako

B Guindo[1*], P Kitha[2], A Keita[1], M Sangaré[1], P Kamaté[1], F. Dembélé[1], L. Dissa[1], R. Dembele[1], D Tounouga[2], Z Diallo[1]

[1]Dermatology Hospital of Bamako (HDB)
[2]University Dermatology and Venerology Center of CNHU – HKM /Cotonou

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***Corresponding author:** Guindo Binta
Dermatology Hospital of Bamako (HDB)

Abstract **Case Report**

Progressive erythrokeratodermia (PEK) is a rare génodermatose associated with disorders of keratinization. Mutations are found in genes encoding connexins 31 and 30.3 mapped to chromosome 1 p34-35. This disease is characterized by fixed hyperkeratotic plaques and transient erythematous plaques. Approximately 50% of affected individuals develop palmoplantar keratoderma. Connexins are components of gap junctions, which are intercellular channels present in almost all tissues, including the skin. A 15-year-old Guinean female, a seamstress, presented with asymptomatic black patches on her feet that had been evolving continuously for 5 years, initially starting on the feet and spreading elsewhere, gradually increasing in size with a transient tingling sensation. Previous treatment involved topical phytotherapy. She had no significant medical history, no similar skin conditions in the family, and no history of consanguinity between her parents. On examination, erythematous and keratotic plaques of variable shapes and sizes were noted on the backs of the hands, the posterior surfaces of the forearms, both knees, the lower thirds of the legs, and the backs of the feet, giving a sock-like appearance to the latter location. The complete blood count, lipid profile, and fasting blood glucose were normal. Histopathology was not performed. The diagnosis of progressive erythrokeratodermia of Gottron was considered based on the age of onset, clinical characteristics, and location of the lesions. The patient was prescribed topical keratolytics, oral Acitretin 10 mg, and psychotherapy for both the patient and her parents. It often constitutes an entity encountered in a syndromic context, highlighting the necessity for surveillance.

Keywords: Erythrokeratodermia, Progressive, Symmetric, Bamako.

INTRODUCTION

Progressive erythrokeratodermia (PEK) is a rare genodermatosis associated with disorders of keratinization [1]. Mutations are found in genes encoding connexins 31 and 30.3 mapped to chromosome 1 p34-35. This disease is characterized by fixed hyperkeratotic plaques and transient erythematous plaques. Approximately 50% of affected individuals develop palmoplantar keratoderma. Connexins are components of gap junctions, which are intercellular channels present in almost all tissues, including the skin [2, 3]. In order to further advance the understanding of this likely heterogeneous condition, Chinese patients from two multiplex families where transmission clearly occurs in an autosomal dominant mode and a sporadic case were analyzed using a fairly classical exome sequencing technique [3]. Treatment of PEK generally involves the use of topical keratolytics and emollients, resulting in some improvement of hyperkeratosis. Low-dose systemic retinoid may be beneficial. New therapies targeting connexin hemichannels and gap junctions may become available in the future [4, 5]. It does not affect the patient's lifespan. However, due to its disfiguring appearance, it can lead to significant psychosocial consequences on the patient's life and their surroundings.

We report a case of Symmetrical Progressive Erythrokeratodermia (SPEK) or Gottron type erythrokeratodermia due to their rarity.

OBSERVATION

A 15-year-old Guinean female, a seamstress, presented with asymptomatic black patches on her feet that had been evolving continuously for 5 years, initially starting on the feet and spreading elsewhere, gradually increasing in size with a transient tingling sensation. Previous treatment involved topical phytotherapy. She had no significant medical history, no similar skin

conditions in the family, and no history of consanguinity between her parents.

On examination, erythematous and keratotic plaques of variable shapes and sizes were noted on the backs of the hands, the posterior surfaces of the forearms, both knees, the lower thirds of the legs, and the backs of the feet, giving a sock-like appearance to the latter location.

Figure 1: Erythematous and scaly plaques resembling socks on the lower thirds of the legs and feet.

Figure 2: Erythematous and scaly plaques on the backs of the hands and knees.

Figure 3: Erythematous and keratotic plaques on the posterior surfaces of the forearms.

The complete blood count, lipid profile, and fasting blood glucose were normal. Histopathology was not performed. The diagnosis of progressive erythrokeratodermia of Gottron was considered based on the age of onset, clinical characteristics, and location of the lesions. The patient was prescribed topical keratolytics, oral Acitretin 10 mg, and psychotherapy for both the patient and her parents. The response to this treatment was favorable, with a decrease in hyperkeratosis.

Argument

EKPS is a rare dermatosis, which means it deserves special attention as it can be stigmatizing, especially in our settings where an unknown diagnosis, particularly if visible, can pose many problems. Numerous previous observations of this disease were in a syndromic context, highlighting the importance of closely monitoring our patient with regular check-ups, either bi-annually or annually [6, 7]. Our observation is particular because hyperkeratosis presents in an unusual manner on certain parts of the body, notably the acral extremities with a sock-like appearance, as well as its localization to almost all pressure areas of the body, explaining the hyperkeratotic component as in all dermatoses with keratinization disorders. The lesions in our patient would have started in early childhood, as evidenced by certain literature data. This disease warrants special attention as it can become stigmatizing. It is essential for clinicians to be well-versed in this genodermatosis and be able to refer patients to specialists for their management, particularly for psychological support [8].

CONCLUSION

EKPS is a rare genodermatosis that deserves to be known as it can be stigmatizing for the patient and their surroundings. It often constitutes an entity encountered in a syndromic context, highlighting the necessity for surveillance**.**

Conflict of Interest: None

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