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Case Report

Hypohidrotic Ectodermal Dysplasia with Palmoplantar Keratoderma: A Case Report

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Abstract

Introduction: Ectodermal dysplasia (ED) is a rare hereditary disorder involving two or more of the ectodermal structures. Palmoplantar keratoderma is a characteristic feature of hidrotic forms of ED. We report a case of Hypohidrotic ectodermal dysplasia associated with palmoplantar keratoderma. ***Case Report:*** A 4-year-old girl presents a chronic palmo-plantar dermatosis that started three years ago. The interrogation finds a history of hospital admission in the 2nd month of life for an unexplained fever. The dermatological examination finds a limited, diffuse erythematous palmo-plantar hyperkeratosis, associated with diffuse alopecia with light-colored brittle and slow-growing hair. The findings also include hypotrichosis and onychodystrophy. The rest of the examination revealed a facial dysmorphism with a light prominent forehead, flattened bridge of the nose, sunken eyes with periorbital hyperpigmentation and photophobia. Other abnormalities include a hypodontia with small teeth. ***Discussion:*** Our observation suggests a hypohidrotic ectodermal dysplasia. It is a genetic disease most often hereditary. The X-linked form or Christ Siemens Touraine syndrome is the most frequently encountered form. A male predominance is noted. Clinically it is characterized by the presence of all or several of the four typical clinical signs of the disease: Anhidrosis or hypohidrosis, dental hyoplasia, hypotrichosis, facial dysmorphism. Palmoplantar keratoderma is rarely associated as it is the case in our patient. The genetic study reveals abnormalities that can be found in the X chromosome for the X-linked forms, and at the level of the chromosomes 2q11-13 or 1q42 for the autosomal forms. ***Conclusion:*** Our observation illustrates the frequent diagnosis delay in this disease. It also emphasizes the importance of a detailed somatic examination in front of any palmoplantar keratoderma presented in children.

Keywords: Ectodermal dysplasia, Hypohidrotic, Palmoplantar keratoderma.

INTRODUCTION

Ectodermal dysplasia is a rare hereditary disorder involving two or more of the ectodermal structures, which include skin, teeth, nails, hair and sweat glands [1]. Palmoplantar keratoderma is a characteristic feature of hidrotic forms of ED [2]. We report a rare case of Hypohidrotic ectodermal dysplasia associated with palmoplantar keratoderma.

Case Report

A 4-year-old girl, of non-consanguineous parents, is brought by her parents for a chronic palmo-plantar dermatosis that started three years ago.

The interrogation finds a history of hospital admission in the 2nd month of life for an unexplained fever. There are no psychomotor development abnormalities or similar cases in the family.

The dermatological examination finds a limited, diffuse erythematous palmo-plantar hyperkeratosis (Figure-1), associated with diffuse alopecia with light-colored brittle and slow-growing hair. The findings also include hypotrichosis and onychodystrophy. The rest of the examination revealed a facial dysmorphism with a light prominent forehead, flattened bridge of the nose, sunken eyes with periorbital hyperpigmentation and photophobia. Other abnormalities include a hypodontia with small teeth (Figure-2).

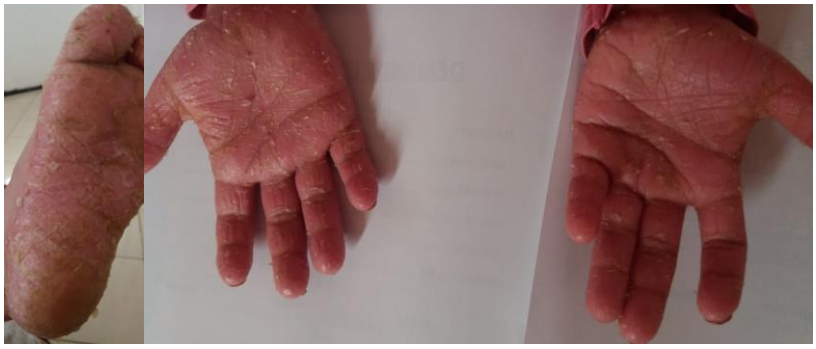

Fig-1

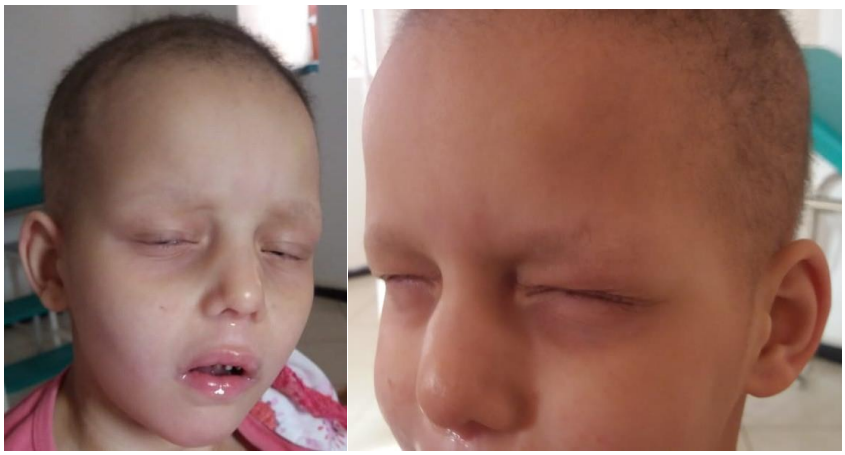

Fig-2

DISCUSSION

Our observation suggests a hypohidrotic ectodermal dysplasia. It is a genetic disease most often hereditary. The X-linked form or Christ Siemens Touraine syndrome is the most frequently encountered form. Its incidence is estimated to occur in 1 in 17,000 newborns worldwide [1]. This condition results from mutations in one of several genes .The most common genetic mutation concerns the EDA gene that encodes the transmembrane protein ectodysplasin-1 (EDA1), a member of the TNFα-related signaling pathway. A male predominance is noted. The identification of female cases is explained by the involvement of both copies of the X chromosome or the existence of different autosomal inheritance patterns whether dominant or recessive [3].

Clinically it is characterized by the presence of all or several of the four typical clinical signs of the disease: Anhidrosis or hypohidrosis, dental hyoplasia, hypotrichosis, facial dysmorphism. In newborns, hypohidrosis is a source of thermoregulation disorders causing unexplained fevers. Palmoplantar keratoderma is rarely associated as it is the case in our patient. The diagnosis is assisted by radiology, which confirms hypoplasia and dental abnormalities, and by histology, which shows rare and hypotrophic sweat glands [2]. The genetic study reveals abnormalities that can be found in the X chromosome for the X-linked forms, and at the level of the chromosomes 2q11-13 or 1q42 for the autosomal forms [1].

Patient's care is based on general measures against hypohidrosis consequences and local treatments for cutaneous manifestations. Global maxillofacial management will seek to better the masticatory function, the growth of the facial morphology and the psychological comfort [2].

Our observation illustrates the frequent diagnosis delay in this disease and the existence of female forms with a marked clinical presentation. It also emphasizes the importance of a detailed somatic examination in front of any palmoplantar keratoderma presented in children.

CONCLUSION

Christ-Siemens-Touraine syndrome is a genetically and a phenotypically heterogeneous disease. It is important to recognize it early to avoid the complications of hypohidrosis and start multidisciplinary care. The family survey will determine whether it is a family form or a sporadic case.

Conflict of Interest: The Authors declare no conflict of interest.

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