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A CASE REPORT OF HYPOHIDROTIC ECTODERMAL DYSPLASIA

Dr. M. Manikandan* Final year postgraduate, Sri Venkateswara Medical College, DR. NTR University of Health Sciences. *Corresponding Author

Dr. Balaji Kakarla Final year postgraduate, Sri Venkateswara Medical College, DR. NTR University of Health Sciences.

ABSTRACT Ectodermal dysplasias, a group of inherited disorders, are characterized by developmental abnormalities of two or more ectodermal structures, one of which at least involves the hair, teeth, nails or sweat glands and other ectodermal structures. Hereby, we report a case of Hypohidrotic Ectodermal Dysplasia.

KEYWORDS : Hypohidrotic, Ectodermal Dysplasia, Hypotrichosis, Hypodontia

INTRODUCTION

Ectodermal dysplasias, a group of inherited disorders, are characterized by developmental abnormalities of two or more ectodermal structures, one of which at least involves the hair, teeth, nails or sweat glands and other ectodermal structures. X-linked Hypohidrotic Ectodermal Dysplasia (HED) is the most common of the ectodermal dysplasia, comprises of hypotrichosis on the scalp but also of the eyebrows and eyelashes, hyperpigmentation around the eyes, dry skin and peg shaped primary and secondary teeth with hypodontia. We are here reporting a classical case of 2 year old boy with HED.

CASE REPORT

A 2 year old boy, born out of second degree consanguinity, by normal vaginal delivery, at term without any complications, brought by his mother to us with complaints of absence of hair all over the body, decreased sweating and heat intolerance since birth. There is no family history of similar illness.

On cutaneous examination, scalp hair was scanty, light-colored, thin with total absence of eyebrows and eyelashes. There was hyperpigmentation around eyes, nose and mouth. The skin all over the body was dry and wrinkled. The examination of oral cavity showed wide-spaced, peg shaped upper incisor teeth and hypodontia. The skin over palms and soles was dry and thickened. The nasal bridge was depressed and nails were normal. Routine general and physical examination were normal. A clinical diagnosis of Hypohidrotic Ectodermal Dysplasia (HED) was made based on clinical history and examination findings.

Figure 1: Photograph Of Child Showing Sparse Scalp Hair, Complete Loss Of Eyebrows And Hyperpigmentation Around Eyes, Nose And Mouth.

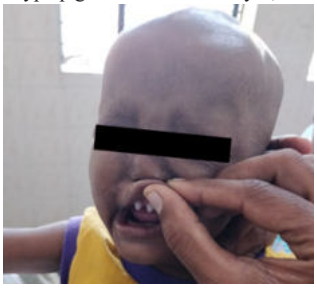

Figure 2: Photograph Of Child Showing Conical Or Peg Shaped, Wide-spaced, Upper Incisor Teeth.

DISCUSSION

Ectodermal dysplasias are a group of disorders characterized by defects in the hair, nails, teeth, and sweat glands. This disorder is classified in several ways. Friere - Maia and Pinheiro suggested a classification depending on the ectodermal derivatives involved. (1) This classification contains more than ten subgroups.

HED can be inherited in three ways: X-linked recessive, autosomal dominant, or autosomal recessive. The X-linked recessive inheritance is found in 95% of randomly selected people with HED. The rest (5%) are either autosomal recessive or autosomal dominant. Mutations in the genes encoding several proteins involved in the ectodysplasin signal transduction pathway cause HED. The most common form, X linked hypohidrotic ED, is characterized by a group of hair and tooth anomalies as well as an decreased sweating. (2) The affected neonates have a collodion-like membrane as well as pronounced scaling. With light-brown coloration, the scalp hair is scanty or completely lacking as seen in this present case. Affected children present clinically with unidentified pyrexia and hyperthermia as early as the first few hours of life. This is caused by the inability to sweat to a noticeable degree, which causes an increase in the body's core temperature. Overall, a history of heat intolerance is significant but not compulsory as seem in this present case. The skin appears smooth due to dermatoglyphic disruption caused by the lack of eccrine pores. A characteristic feature is facial dysmorphism.

The conical and pointed teeth are important characteristics of the syndrome and may be the only apparent abnormality. Typically, the incisors and/or canines are affected as seen in this present case. (3) Nails were normal, in comparison to various other types of ectodermal dysplasia. Scaling or plucking of the skin during the neonatal period, periorbital hyperpigmentation as seen in this present case and wrinkles, facial sebaceous hyperplasia, and eczematous dermatitis are other cutaneous features of HED.

The management of children and adults with HED is difficult due to their heat intolerance (particularly during febrile illness or physical exercises in hot conditions) and vulnerability to pulmonary infections. During hot temperatures, people with the condition must have access to a sufficient supply of water as well as a cool surroundings, which may include "cooling vests," air conditioning, a wet T-shirt, and/or a water spray bottle. External cooling, however, is less effective in these patients because heat transfer from the core to the skin is also reduced, probably due to inadequate capillary dilatation. (4)

CONCLUSIONS

Ectodermal dysplasias (EDs) are rare genetic skin diseases with numerous variations that include ectodermal structures. Because patients are usually children, parents should be instructed about the disease and its symptoms if there is a family history of similar conditions. Parents must also be reassured that their children can live a normal life if they are diligent about regular checkups and seek medical attention immediately if any symptoms arise.

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