Hypohydrotic Ectodermal Dysplasia in Black Africans

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ABSTRACT

Ectodermal dysplasia is an uncommon genodermatosis which may be associated with mental retardation. The diagnosis is often missed in newborns or delayed due to its varied overlapping clinical presentation in affected individuals. We report two maternal blood related males with complaints of heat intolerance since birth, lack of sweating and dry scaly skin. Both had normal developmental milestone and mental acuity. The treatment is symptomatic and patient dependant while overall management requires a multidisciplinary team approach.

Key words: *Hypohidrotic, ectodermal dysplasia, hypodontia*

Siyah Afrikalılarda Hipohidrotik Ektodermal Displazi

ÖZET

Ektodermal displazi mental retardasyon ile ilişkili nadir genodermatozisdir. Tanı sıklıkla yenidoğanlarda atlanılır ve kişileri etkiliyen diğer klinik durumlar nedeniyle geç konulur. Biz terleme eksikliği ve kuru deri nedeniyle doğumda ısı intoleransı gelişen 2 erkek hasta bildirdik. Tedavi semptomatik ve multidisipliner bir yaklaşım ile hastaya göre değişmektedir.

Anahtar kelimeler: *Hipohidrotik, ektodermal displazi, hipodonti*

INTRODUCTION

Ectodermal dysplasia (ED) is an inherited disorder of the ectoderm characterized by defective development of the hair, teeth, nail and sweat gland (1). It is classified into four primary defect groups based on the ectodermal structure affected and over 170 clinical types with overlapping features have been identified (1-3). Thus, diagnosis may be missed or overlooked in affected newborn and infants. ED is further categorized into either hidrotic or hypohidrotic (anhidrotic) depending on the primary defects involved and may be associated with mental retardation (4-6). Over a 20 year period spanning 1991-2010, four cases of ectodermal dysplasia were diagnosed in the Department of Pathology, Ahmadu Bello University Teaching Hospital Zaria. We present two of the cases. The developmental histories, clinical presentations and tissue histology from palm biopsies formed the basis of diagnosis. Tissue biopsies were processed in paraffin wax and histology sections were stained with haematoxylin and eosin (H and E).

CASE

A 6year old boy and his 21years old maternal uncle both complained of persistent heat intolerance since birth necessitating frequent drenching with water, lack of sweating and dry scaly skin. Both had normal developmental milestones except for the absence of teeth eruption. Both have normal mental acuity. The uncle is a polytechnic student whose medical challenges were thought to be a one-off incidence by the family members until the boy started showing similar symptoms. Their clinical examinations revealed abnormal facies comprising hypoplastic maxilla and mandible, absent

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***Figure 1**. Frontal bossing, depressed nasal bridge and thin sparse hair*

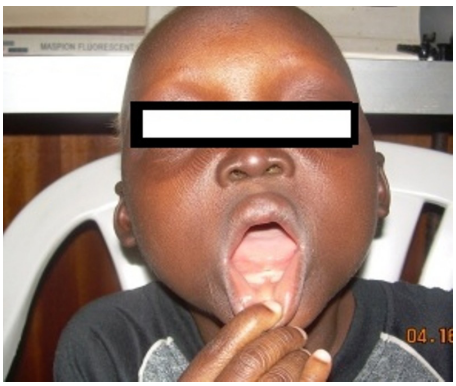

***Figure 2**. Absent teeth, patulous lips and normal nail*

eyebrows, sparse thin silky scalp hair, generalized hyper-pigmentation, patulous lips, normal nails and dry scaly skin (Figure 1). The boy had two hypoplastic pointed incisors in the lower jaw while the uncle had no teeth (Figure 2). The boy's mother and young sister were clinically unaffected.

Tissue histology of the full thickness biopsies from their palms showed hyperkeratotic and acanthotic epidermis with broad elongated rete pegs overlying a densely collagenized dermis lacking skin adnexal structures (Figure 3). A clinico-pathologic diagnosis of hypohidrotic ectodermal dysplasia was made. Both patients absconded after tissue biopsies were taken, and efforts to locate them through their home address were fruitless.

DISCUSSION

Hypohidrotic ectodermal dysplasia (HED) also known as Christ-Siemens- Touraine syndrome and characterized by absence of sweat glands and abnormalities of the hair follicles and teeth was first described by Thurnam (7). Since then, many clinical variants have been reported in literature. The earliest cases seen in Nigeria was reported in a single family in 1975 (8). HED results from mutations of the ectodysplasin A (EDA), EDA receptor (EDAR) and EDAR associated death domain (EDARADD) genes involved in the signal transduction pathway of ectodysplasin which regulates the morphogenesis of skin appendages (9-12). The X-linked EDA is the commonest form and affects males predominantly while EDAR and EDARADD mutations occur as autosomal disease in a smaller percentage of individuals and females are usually heterozygote with varied phenotypic expression (3,10,13). The first cases of ED seen in our hospital and referred to our department were a set of identical female twin with autosomal recessive manifestation (14). However, the X-linked EDA and the autosomal form may sometimes be indistinguishable clinically while the autosomal dominant HED is readily differentiated from autosomal recessive disease clinically (9).

Distinctive attributes of classic X-linked HED include reduced or absent sweating (hypohidrosis), frontal bossing, flat or depressed nasal bridge, hyperpigmented wrinkled skin especially around the eyes, patulous lips, sparse thin scalp and body hair (hypotrichosis), malformed or absent teeth (hypodontia) and bad smelling nasal discharge (ozena) (1,15-17). The nails are usually unaffected and normal. The full hypohidrotic phenotypic expression was seen in both males.

The diagnosis of HED is often missed in newborns or

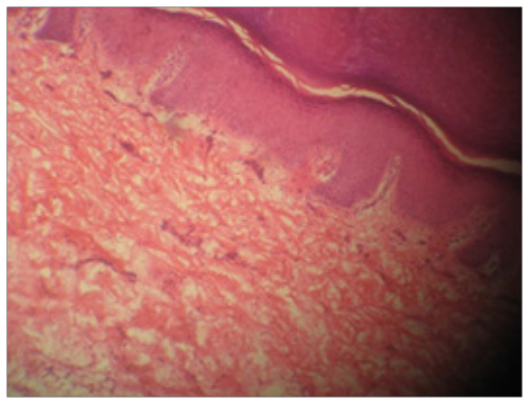

***Figure 3**. Skin biopsy appendage structures*

delayed in infant due to its varied overlapping clinical presentation in affected individuals as seen in these cases. Definitive diagnosis is a combination of clinical features, genetic studies to locate the mutated genes and tissue biopsy taken from the axilla and palm which will show hypoplastic to absent eccrine glands as seen in our cases. It is also important to differentiate the HED facie from the frontal bossing resultant from extra medullary haemopoeisis seen in sickle cell disease especially in our setting and other congenital disorders such as Rothmund- Thomson and Cockyane syndromes and achondroplasia (18).

Treatment is symptomatic while overall management requires a multidisciplinary medical team approach dependant on the age of the patient. Temperature control is paramount especially for our patients who live in hot arid tropical zones. Skin lesions and secondary infections from cracked scaly skin should be treated promptly. Regular denture fittings and prosthesis that will aid development of the jaw bones will also be of benefit to some of these patients. Counseling and screening should be advocated in other members of the family.

In conclusion, ectodermal dysplasia is a heredo-familial disease with a male predilection. It may be inherited either in an autosomal or x-linked pattern. Our cases conformed to the hypohidrotic form of the disease which affected all but one primary ED defects. Its management is dependent on the presentation of affected individual while temperature control is vital to patient's survival in tropical environment.

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