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Case Report

Hypohidrotic ectodermal dysplasia

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Abstract Ectodermal dysplasia (ED); is a rare disorder affecting ectodermal derivatives i.e. skin, nails, teeth, hair and sweat glands. It is divided into many subtypes. Among those, two major types involve (a) Hypohidrotic/ Anhidrotic ED (Christ-Siemens-Touraine Syndrome) and (b) Hidrotic ED (Clouston Syndrome). Here, we present a case of hypohidrotic ED in a 19 years old male as a rare presentation in our region.

Key words
Ectodermal dysplasia; Hypohidrosis; Hypodontia; Hypotrichosis.

Introduction

Ectodermal dysplasia; is a rare entity that mainly affects ectodermal derivatives i.e. skin, nails, hair, teeth and sweat glands. Among ectodermal dysplasias, most common types are hypohidrotic/ anhidrotic ED in which sweat glands are largely affected and hidrotic ED where sweat glands are not affected.[1] The incidence of HED is around 7 cases per 10,000 live births.[2] There are different patterns of inheritance of this disease i.e. X-Linked recessive (XLHED), Autosomal Dominant (ADHED) and Autosomal Recessive (ARHED).[3] This condition is characterized by hypotrichosis, hypodontia and hypohidrosis. Other systemic features include frontal bossing and depressed mid face. There are sparse and slow growing scalp and body hair. Nails are often normal or show onychodysplasia.

Hypohidrotic ED1 is caused by EDA1 gene mutation that encodes ectodysplasin. Most often, females act as carriers of X-linked pattern of disease.[4] This disease is associated with asthma, atopic eczema and recurrent pneumonia. Hypohidrosis can lead to life threatening heat intolerance. About 21% of XLHED cases reported a strong family history of infant or early childhood deaths.[5]

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Case report

A 19 years old male presented to Dermatology Department of Allama Iqbal Memorial Teaching Hospital, Sialkot having history of sparse, thin scalp and body hair along with absent eyebrows. Teeth were missing since childhood. On examination, he had frontal bossing, broad nose, periorbital skin wrinkling and increased pigmentation since young age **(Figure 1)**. His nails were normal **(Figure 2)**. Our patient complained about decreased sweating which was very troublesome for him. He had to combat with his increased body temperature by repeated cooling of body with water, drinking cold beverages frequently and living in air conditioned places. He had multiple visits to the hospital because of recurrent bouts of hyperthermia.

Interestingly, he reported that two of his elder brothers were having the same cutaneous

Figue 1

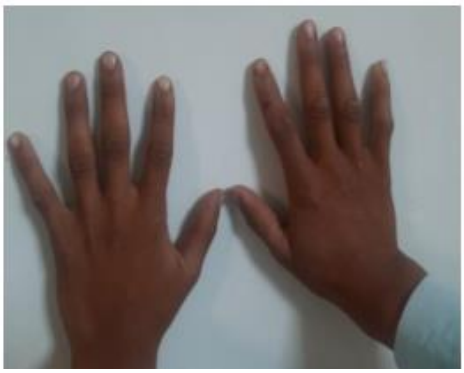

Figure 2

Figure 3

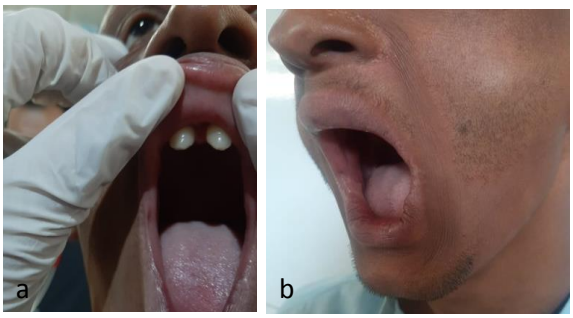

Figure 4

features and severe heat intolerance due to loss of sweating which led to the death of one brother at the age of 3 years and the other one at 2.5 years. Further now, his nephew who is 9 months of age was developing similar condition **(Figure 3)**.

His biochemical profile, including complete blood count, liver function tests, renal function tests, urine analysis, and ultrasound abdomen were normal. Upon intraoral examination, his teeth were missing **(Figure 4b)** and the present ones were widely spaced and peg shaped **(Figure 4a)**. So, on the basis of these clinical features we diagnosed this as a case of HED.

Discussion

Ectodermal Dysplasia, is caused by a genetic mutation in ectodermal dysplasin A gene. It can be broadly classified into two major groups; hypohidrotic ectodermal dysplasia and hidrotic ectodermal dysplasia. The most commonly seen form is X-linked hypohidrotic ectodermal dysplasia that is characterized by hypohidrosis, hypodontia/ adontia and hypotrichosis (Christ-Siemens-Touraine Syndrome).[6]

Facial features include frontal bossing, low set or spock ears, broad/ saddle nose, periorbital wrinkling and hyperpigmentation with sparse or absent eyebrows. Scalp and body hair are hypopigmented, thin and lusterless. Nails can be normal or show onychodysplasia.[3]

Major cutaneous feature of this disease is decreased sweating leading to severe heat intolerance, therefore they often prefer cool environment, plenty of cool drinks and wet clothes. This makes the summer months very stressful for the affected ones. Hypotrichosis and sparse body hair makes the physical appearance unacceptable for the patient leading to social reluctance.

Intraoral features such as hypodontia/ adontia and conical teeth can be seen in these patients. This orthodontic problem makes chewing difficult for them and can lead to low self esteem. HED also involves mucous glands resulting in thick secretions that can cause recurrent respiratory infections.[7]

Other subtypes of hypohidrotic ectodermal dysplasia including Ectrodactyly Ectodermal

Dysplasia Cleft lip/palate (EEC syndrome) and Ankyloblepheron, ectodermal defects, cleft lip/ palate (AEC syndrome) also share similar clinical features. EEC syndrome is characterized by abnormalities of hairs, nails and teeth with additional findings of ectrodactyly, lacrimal duct anomalies, cleft lip/palate and renal abnormalities. Comparably, patients of AEC syndrome present with ankyloblepharon (partial thickness fusion of eyelid margins) along with hypotrichosis, hypodontia, dystrophic or absent nails. These above mentioned features of EEC and AEC syndromes were absent in our case.

X-linked hypohidrotic ED with immunodeficiency features abnormal hair growth, pyrexia of unknown origin, malformed or absent teeth with vascular anamolies and recurrent cutaneous and systemic infections. Our patient had no history of repeated systemic infections with cutaneous manifestations. So, this comprehensively favours our diagnosis of HED.

Dermatopathia pigmentosa reticularis, which is an autosomal dominant ectodermal dysplasia that shows poikiloderma and reticulate hyperpigmentation, non-cicatricial alopecia and onychodystrophy alongwith hypotrichosis, hypohidrosis and poorly developed teeth. Due to the absence of poikilodermatous changes and reticular pattern of pigmentation in our patient, hypohidrotic ectodermal dysplasia was the favoured diagnosis.

Conclusion

Hypohidrotic Ectodermal Dysplasia is a rare disorder. It can lead to low self-esteem and social isolation. To improve the quality of life for these patients early orthodontic procedures/implants are worth considering. For hair problems topical minoxidil, hair transplant and wigs can be beneficial for them. Genetic counselors play a pivotal role in counseling and reassurance.

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