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Management of Skin Symptoms in a Rare Case of Hypohidrotic Ectodermal Dysplasia

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ABSTRACT

Hypohidrotic ectodermal dysplasia (HED) is a rare genetic disorder that affects multiple body systems and is characterized by a triad of symptoms that impact hair, teeth, and sweat glands. Diagnosis of HED is primarily based on physical features and is confirmed through genetic testing. Currently, the available treatment options for HED are limited to general medical interventions such as the use of skincare products and prosthetic dental treatment. We present a patient with HED who exhibited typical symptoms in the face and oral cavity. The patient was treated with a topical combination therapy for severe dyskeratosis. The purpose of this report is to increase awareness of HED within the dermatology community and provide information on its diagnosis and management also early diagnosis can enable prompt intervention, relevant therapy and support.

INTRODUCTION

Hypohidrotic ectodermal dysplasia (HED) is a rare genetic disorder that affects the development of ectodermal structures like hair, teeth, nails, and sweat glands (1, 2). HED is typically inherited in an X-linked recessive pattern and is caused by mutations in the EDA gene (3). The main symptoms of HED include reduced ability to sweat, abnormal or missing teeth and sparse hair. Severe cases may also experience febrile seizures and issues with thermoregulation (3-5).

Diagnosis of HED is based on physical features and confirmed through genetic testing. While there is currently no definitive cure for HED, general medical interventions like avoiding overheating, using skincare products and prosthetic dental treatment can help manage symptoms (5, 6). To date, no gene therapy for HED has been reported in humans (5). However, ongoing research and advancements in the field hold the promise of new treatments and diagnostic tools in the future.

CASE REPORT

A 4-year-old and 5-month boy was referred to the pediatric department of Honary Clinic in Jahrome, Iran, with complaints of lethargy, weakness and heat intolerance after sun exposure. During the physical examination, it was noted that he had wrinkled, hyperpigmented skin around his eyes with very scanty eyelashes and eyebrows. His scalp hair was light, thin, and sparse, and he has been suffering eyelash loss as well, particularly in the summer (Figure 1). In the oral examination, the child had malformed teeth and began dentition at 1 year old, with fewer and conical- shaped teeth. His hypodontia was asymmetrical, with one tooth on the mandible and six on the maxilla. The ear implantation was normal, but he had micrognathia and a depressed nasal bridge. Upon examination of his extremities, the skin was noted to be dry and wrinkled, and severe bilateral plantar dyskeratosis was detected. Although his nails were normal with no signs of dystrophy, his toenails were found to be fragile. His neurological development was normal, and he had speaking problem. However, he had difficulty in eating

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including chewing solid food and addition, his weight was less than average for his age (12.5 Kg 5-10% weight-for-age, 99 cm 10-25% height-for-age). His past medical history revealed that, he had an inguinal hernia. The child was born to a non- familial intermarriage, with a Tajik mother and an Arab father. He is the fifth child of his family. His mother had a mild presentation of HED; whose features are consistent with hypodontia, heat intolerance and dry skin but no evaluation was done due her low socioeconomic status.

INVESTIGATION

To rule out other possible diagnoses, echocardiography and abdominopelvic sonography were performed for the patient, and the results were normal. The child was also assessed by a pediatric gastroenterologist due to the dysphagia and the low percentile of growth; no other pathology was found (Table 1).

The hypodontia was also confirmed by panoramic radiography imaging.

Table 1. Laboratory tests at presentation

Test	Result	Unit	Normal Range
Total leukocyte count	7.0	X10^3/Mic	4.4-11.3 :10^3/micl
Hemoglobulin	13.0	g/dl	14-18
Hct	35.7	%	41-50
MCV	74.8	F lit	80-96
MCHC	36.4	g/dl	31-36
Platelet	449	X10^3/Mic	150-450
BUN	13	mg/dL	8-20
Cr	0.5	mg/dL	0.9-1.3
TSH	0.62	mic Iu/ml	0.39-6.16
T4	7.17	Mic g/mL	4.5-12.5
T3	1.03	Ng/ml	0.52-1.85
Vitamin D3	53.4	Ng/ml	Sufficiency:30-100
Ferritin	67.7	Ng/ml	16-220
Fe	95	U/ML	40-168
TIBC	194	Mic g/ml	250-450
Fasting blood sugar	70	Mg/dl	1-6 years 74-127
HbA1C	4.9	%	3-6%
Chol	151	mg/dL	<200
TG	51	mg/dL	<200
LDL	101.8	mg/dL	<130
HDL	39	mg/dL	30-70
Bili.T	0.49	mg/dL	0.1-1.2
SGOT	38	U/L	<38
SGPT	16	U/L	<41
Alk.pho	321	U/L	100-460
T.pro	7.1	g/dl	4.4-10
Alb	3.9	g/dl	3.5-5.2
Ca	9.1	mg/dL	8.6-10.3
Phos	4.9	mg/dL	3-5.4
Urine culture	Negative	Qualititavive	Qualititavive
Urine analysis	Negative	Qualititavive	Qualititavive
PH	7.38	mmHg	7.35-7.45
PCO2	35.5	mmHg	35-48
HCO3	21.1	mmHg	18-23
Wright	Negative	Qualititavive	Qualititavive
2ME	Negative	Qualititavive	Qualititavive
Anti-TTG	Negative		
IgA total	Normal		
Sonography	Normal		
Echocardiography	Normal		

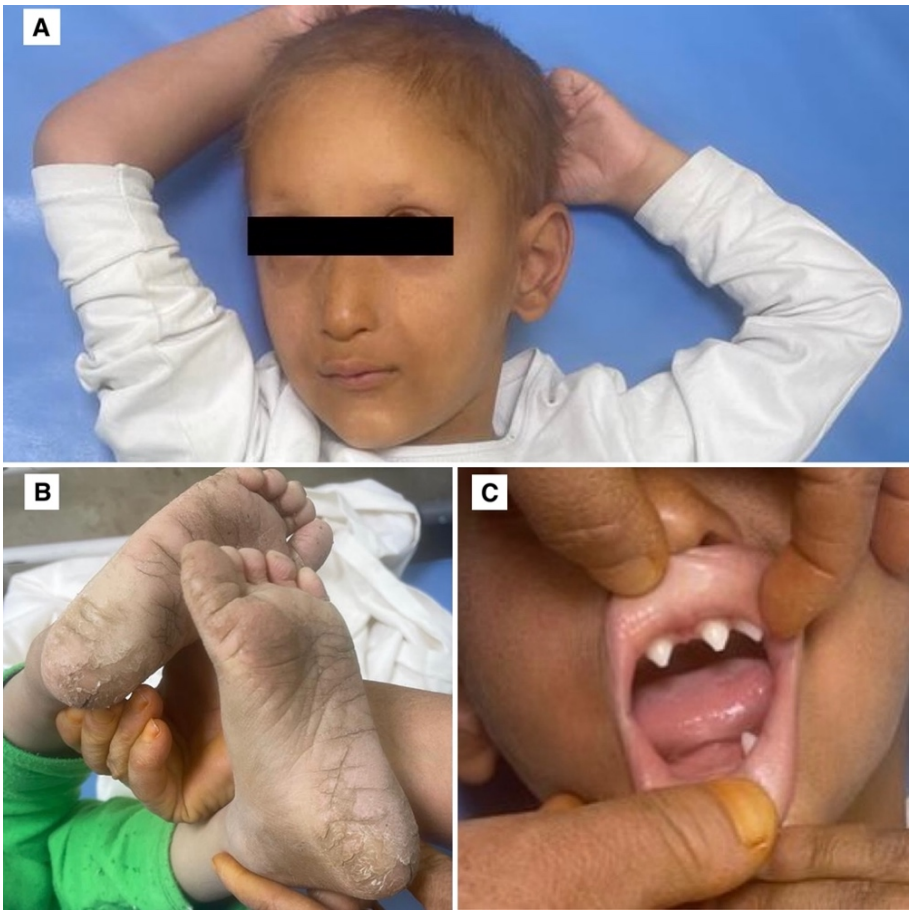

FIGURE 1. (A); wrinkled, hyperpigmented skin around his eyes with scanty eyelashes and eyebrows. **(B);** Severe plantar dyskeratosis is present. **(C);** Teeth are reduced in number and are conical in shape.

SKIN BIOPSY

A skin biopsy was not performed due to clinical evidence in the patient.

DIFFERENTIAL DIAGNOSIS

Non- specific partial anodontia, syndromic partial anodontia and trichodento- osseous syndrome.

TREATMENT

The patient's parents were educated about the required lifestyle and warned against sun exposure. Maintain cool surrounding temperature with air conditioning, light clothing, plenty of fluids. Consuming sun cream was advised. It was also recommended to increase fluid intake and use soft diet.

For the relief of skin symptom, the combine hydrocortisone emollient was prescribed with formulation of 20% Megacort (mometasone furoate) + 5% dexpanthenol + 5% urea in a 70% eucerin use every night for 1 week, then 2-3 time in a week.

OUT COME and FOLLOW UP

The patient was kept under follow-up for six months. Due to the patient's young age and micrognathia, prosthetic treatment had not yet been initiated.

DISCUSSION

HED is a heterogenous group of disorders that share the following features; heat intolerance and fever due to partial or complete absence of sweat gland, missing or malforms teeth, spare and hypopigmented hair (1). HED also have facial feature like; depress nasal bridge, frontal bossing, micrognathia and malar hypoplasia (6).

So far, more than 192 distinct disorders have been described. the incidence of HED is estimated to be 1 per 100,000 births, with the severity of symptoms varying between and within families and the most common form of HED is X-linked hypohidrotic ectodermal dysplasia (XLHED) (3). As genetic testing was not performed on the patient, we were unable to identify the specific gene involved in their case. EDA1 is a membrane- bound protein that plays a role in the signaling pathway related to TNFα, facilitating the attachment of ectoderm to the mesenchyme. Its 0importance lies in the formation of hair, teeth, skin and sweat glands (5).

HED management needs a multidisciplinary treatment approach so we referred our patient to a specialist pediatrician, orthodontist, dermatologist, psychiatrist and genetic counseling.

Treating dry skin and atopic dermatitis in individuals with HED involves employing moisturizers, emollients and topical treatments to relieve symptoms. Regular skincare, including gentle cleansing, was advised, also consuming sun cream was advised (7).

In this particular case, a topical combination therapy was administered to address the severe skin symptoms and hyperkeratosis, as mentioned earlier. The specific details of this combination therapy were not documented in other HED case reports. However, given the observed improvement and reduction in the severity of symptoms in the patient, this combination therapy could be considered as a potential treatment option for other HED patients who exhibit similar skin manifestations.

CONCLUSION

We're presenting a case report on a toddler patient who was diagnosed with HED. Our objective is to emphasize the importance of early diagnosis, advocate for a multidisciplinary approach for the management. Our goal is to increase awareness of this condition, enhance patient outcomes for newborns and those already diagnosed with HED, and improve understanding of this rare genetic disorder. It's essential to raise awareness about HED, offer support to affected individuals and their families, and begin management as soon as possible to improve their quality of life.

DECLARATIONS

The authors declare that they have no competing interests. The authors received no specific funding for this work. The authors do not hold any stocks or shares in any organization that may benefit or suffer from the publication of this paper.

AUTHORS' CONTRIBUTIONS

All authors contributed to this project and article equally. All authors read and approved the final manuscript.

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CONFLICT OF INTEREST

Authors declared any conflicts of interest.

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