

Hypohidrotic Ectodermal Dysplasia: A Case Report

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ABSTRACT

Ectodermal dysplasia is a heterogeneous group of inherited disorders that manifests a variety of deformities due to primary defects in the development of two or more tissues derived from embryonic ectoderm involving skin, sweat glands, hair, nails, and teeth. The two most common forms of disease are Hypohidrotic/Anhidrotic Ectodermal Dysplasia and Hidrotic Ectodermal Dysplasia caused by mutation of several genes. Here we present a case of a 4-and-half-year-old male child with Hypohidrotic Ectodermal Dysplasia presented with raised body temperature, heat intolerance, hypodontia, and hypotrichosis. A combined approach with physicians from several fields brought us to a final diagnosis of Ectodermal dysplasia. Proper counseling for parents is essential to ensure timely and comprehensive medical care when needed in the future.

Keywords: *ectodermal; hydrotrichosis; defects.*

INTRODUCTION

Ectodermal dysplasia (ED) is a rare hereditary disorder characterized by abnormal primary development of tissues derived from ectoderm including skin, hair, nails, teeth, and eccrine glands.¹ Clinical features includes fine blond and scanty hair, reduced number of eyelashes and eyebrows, fine and smooth skin with deficient sweat and sebaceous gland, saddle nose, periorbital pigmentation.² The incidence of ectodermal dysplasia is around 7 in 10,000 live births with Hypohidrotic ED being the most prevalent type.³⁻⁵ This disorder can result from a mutation in genes such as ectodysplasin A (EDA), EDA receptors (EDAR), EDAR-associated death domain (EDARADD), and WNT signaling pathway, Indiana Optometric Association (WNT IOA) genes, among which EDA gene mutation accounts for more than half of all cases.⁶ Here we document a 4-and-a-half-year-old boy with Hypohidrotic ED concerning primary and permanent dentition and absence of sweat gland leading to excessive heat intolerance.

temperature, decreased sweating, dry skin, and delayed eruption of abnormally shaped teeth. These symptoms became more prominent causing significant distress to the patient during outdoor physical activities, relieved only with cooling techniques like frequent cold-water showers. The parents denied having a family member with similar features as their child. His birth history was insignificant.

A thorough developmental history of this child revealed developmental delay, mostly pronounced in motor components of developmental milestones. The patient was conscious and cooperative with syndromic facies. On examination, the patient had frontal bossing, sparse hypopigmented hair with a lusterless appearance over the scalp, hyperpigmented skin around the eyes and over the nose, and an absence of eyebrows and eyelashes (Figure 1A,1B).

CASE REPORT

A 4-and-a-half-year-old male child was brought to our pediatric OPD by his parents with complaints of raised body



Figure 1A. Tense bulging skull with no eyebrows and eyelashes and depressed nasal bridge **1B.** Sparse hypopigmented hair

There was decreased lubrication and tearing in both eyes, and there were no vision or hearing issues. The nasal bridge was depressed/flat with normal set ears (Figure 1A). The upper lip was slightly everted with dry oral mucosal membrane. Upon intraoral examination, tongue tie was noted with only a few conical-shaped teeth (pegged teeth) with wide spacing over the upper jaw, and no teeth eruption over the lower jaw (Figure 2).



Figure 2. Pegged-shaped teeth

Systemic examinations otherwise showed normal findings except undescended testes.

The patient was then admitted and laboratory investigations including a complete blood count, comprehensive metabolic panel, TFTs, and urine analysis were sent which reports were all within the normal range. X-rays of long bones and joints revealed a normal bone age of around 4 years old child. Further dermatology, ophthalmology, and dental evaluation were done and managed accordingly. A multidisciplinary approach with detailed history, clinical presentations, and thorough examination played a key role in promptly diagnosing this condition and excluding other differential diagnoses such as multivitamin deficiency and chronic protein-energy malnutrition. As for the treatment of this patient, multivitamins, and moisturizing cream for dry skin were prescribed, and oral hygiene instruction was enforced with periodic dental consultation for further management.

The patient's general condition improved at the 1-month follow-up, and dental evaluation was done on the same visit.

DISCUSSION

Ectodermal dysplasia is a hereditary disorder that affects the growth and homeostasis of two or more ectodermal derivatives including hair, teeth, nails, and special glands,^{5,7} all of which, except nails, were affected in our patient. Depending on the presence or absence of sweat glands, the disease is broadly classified into hypohidrotic/anhidrotic (Christ–Siemens–Touraine syndrome) or hidrotic (Clouston syndrome).^{6,8,9} Our patient is a case of Hypohidrotic ED (HED) which is the most common type of ED; inherited as an X-linked recessive or autosomal dominant trait.^{5,10}

The earliest recorded cases of ED were described in 1792,¹¹ although Thurnam published the first report of patients with ED in 1848.⁵ However, the term Ectodermal Dysplasia was not coined until 1929 by Weech.^{10,12}

ED occurs due to the defects of the genes like ED-1, muscle segment homeobox homology-1, ectodysplasia-1 receptor (EDAR), paired box genes, and human homolog of mouse dl, EDARDD (EDAR death domain gene), and WNT/catenin pathway mutation. The EDAR death domain (EDARDD) gene acts through the EDAR signaling pathway, which binds to intracellular molecules to activate nuclear factor (NF-κB).¹³ Also, the WNT/catenin pathway consists of proteins that play a vital role in embryonic development and bone marrow stromal cells (BMSCs).¹⁴ Mutation of the EDARDD gene or EDA receptor and the WNT/β-catenin pathway leads to the downregulation of NF-κB and osteogenesis. An altered epithelial-mesenchymal interaction is one of the most important pathogenesises of ED.¹³ Since genetic testing was not done in our patient, given the clinically evident features and lower socio-eco-

nomie conditions, we do not know the genes affected.

The triad of hypodontia or anodontia, hypotrichosis, and hypohidrosis is usually seen in Ectodermal Dysplasia,⁶ which were clinically evident in our case. Our patient presented with dry skin, decreased sweating, pegged teeth with delayed eruption, a tense bulging skull, absent eyebrows, sparse hypopigmented hair, and a saddle nose. Patients with ectodermal dysplasia usually have a normal lifespan.⁹ However, complications such as failure to thrive, lung infections, and hyperthermia, may lead to mortality in children, which can reach 30% in the first three years of life.⁷

The patient is diagnosed via thorough history and clinical examination which, however, may be difficult in newborns as features usually become apparent during infancy or childhood. In our patient, skin excoriations in hands and feet were the earliest manifestations noted after 2 weeks of delivery. Family history was insignificant in our case, which may otherwise be helpful in the case of neonates for similar clinical features in the family. Other investigative procedures that aid in diagnosis are radiographs, sweat pore counts that can be easily performed using starch iodine test, pilocarpine iontophoresis, skin biopsy, and genetic testing.¹⁰

A multidisciplinary team of psychiatrists, dermatologists, speech therapists, oral surgeons, prosthodontists, orthodontists, and pedodontists is necessary for the treatment of ED.¹⁰ The treatment is symptomatic and involves protection from heat exposure and care of skin, hair, ear, nose, and nails.⁸ Heat exposure was managed using physical cooling techniques like wearing special cooling vests to reduce heat generation during activities. For all patients with hypohidrotic ED, a dental referral is neces-

sary, so a dental consultation for our patient was done. Implants in children are generally not advised since the mandible and maxilla continue to develop until adolescence. However, some literature supports that implant-supported dentures can be a reliable alternative for children.¹⁶ Similarly, artificial tears and saliva may be helpful for reduced lacrimation and xerostomia, respectively. There are no dietary restrictions indicated in ED.¹ Dermatology consultation was done in our patient for skin excoriations. The patient was prescribed multivitamins and moisturizing creams. Also, proper oral hygiene instruction and counseling were done.

Ectodermal dysplasia is preventable to some extent through proper genetic counseling and avoidance of consanguineous marriages. It is also of paramount importance in the medical field to ensure medical professionals know this condition as it can be diagnosed clinically without undergoing further extended diagnostic approaches. It has an excellent prognosis if precise and effective treatment is given in time. Psychological motivation is just as important for patients to withstand the social stigma associated with early senility.

Consent: Written informed consent was obtained from the patient (or the patient's legal guardian) for the publication of this case report and any accompanying images. Patient anonymity maintained and identifying information has been omitted to the best possible extent to protect the patient's privacy.

Conflict of Interest: None.

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