

CASE REPORT

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Received: 19.07.2024

Accepted: 30.10.2024

Published: 20.12.2024

Citation: Hariharasubramanian M, Rajashekar TS. A Rare Case of Hypohidrotic Ectodermal Dysplasia. J Clin Biomed Sci 2024; 14(4): 167-169. <https://doi.org/10.58739/jcbs/v14i4.69>

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Funding: None

Competing Interests: None

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Published By Sri Devaraj Urs Academy of Higher Education, Kolar, Karnataka

ISSN

Print: 2231-4180

Electronic: 2319-2453



A Rare Case of Hypohidrotic Ectodermal Dysplasia

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Abstract

A wide range of hereditary diseases affecting two or more ectodermally derived tissues together are referred to as ectodermal dysplasias (EDs). The most frequently impacted ectodermal derivatives are the teeth, nails, sweat glands, and hair. The mucous membranes of the mouth and nose, lips, eyes, ears, and other ectodermal structures could also be impacted. During embryonic development, the ectoderm forms the outermost layer of the primary germ layers that give rise to the several structures that are commonly affected in ED. As a result, depending on the array and severity of the anomaly, ED presents itself differently in each patient. This symptom is present in 1 in 50,000 people worldwide. Among these 150 unique syndromes, hypohidrotic (faulty sweat glands) and hidrotic (normal sweat glands) syndromes are the most prevalent. Moreover, there are many inheritance patterns associated with ED, with X-linked inheritance being by far the most prevalent. Here, we report on a seven-year-old boy's clinical case of hypohidrotic (anhidrotic) ED.

Keywords: Hypo Hidrotic Ectodermal Dysplasia; Hair; Skin; Oral Cavity; Xerosis; Hypodontia

1 Introduction

Hypo hidrotic Ectodermal Dysplasia is a genetic skin disease which is inherited in an X - linked recessive, Autosomal Recessive (or) Autosomal Dominant pattern. Common symptoms include sparse scalp & body hair reduced ability to sweat and missing teeth. The incidence of this disease globally is 1 in 50,000 population.¹

2 Case Report

2.1 Clinical History

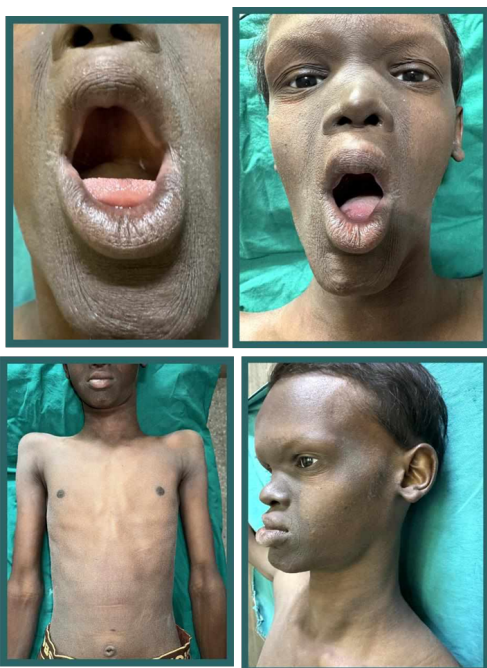
A 14-year-old boy presented with complaints of reduced sweating, sparse hair growth over the scalp, absence of teeth, dryness of skin since birth. Complaints of heat intolerance present during summer, exercise and having spicy food.

History of visible nasal deformity present since birth. History of similar complaints in the family present (Patient had a sibling with similar presentation who passed away at the age of 2).

2.2 On Examination

- **Hair:** Thin light-coloured hair noted over the scalp. Complete absence of eyebrow noted. Presence of pubic and axillary hair noted. Absence of body hair observed.
- **Skin:** Generalised Xerosis present. Periorbital, perioral, and nasolabial hyperpigmentation present. Periorbital wrinkling of skin present. Velvety hyper pigmented skin noted over the axilla.
- **Oral Cavity :** Two molars present in the upper jaw and lower jaw respectively. All other teeth absent. High arched palate present.
- **Nose:** Saddle Nose Deformity noted.
- **Genitalia:** Normal.

Hence diagnosed as Hypo hidrotic Ectodermal Dysplasia.



3 Discussion

A suspected diagnosis of Hypo hidrotic Ectodermal Dysplasia should be accompanied by a thorough history and physical examination. A family history is important in identifying

familial cases and should focus on pigment abnormalities, premature graying of the hair, hearing loss, and gastrointestinal complications. A detailed physical examination should be performed with particular attention to the hair, skin, oral cavity & nasal cavity.

ED is a broad category of genetic illnesses marked by birth abnormalities of one or more ectodermal structures, such as appendages of the skin. All mendelian mechanisms of inheritance have been observed, and the prevalence of ED is estimated to be seven per 10,000 births.² Pregnancy may cause the disease to manifest in the first trimester. If it is severe, the dentition will be impacted since it manifests prior to the sixth week of embryonic life. It's possible for other ectodermal structures to be impacted after the eighth week. Assessing relatives of patients with hypo hidrotic ED and identifying partial form carriers in their families are crucial steps in evaluating intrafamilial genetic transmission. Assume that none of the relatives had experienced any other episodes of ED. In such scenario, it suggests that, as some earlier research has indicated, the proband was most likely brought on by a novel mutation or gene translocation.³

It is important to counsel and advise parents to keep their kids away from physically demanding activities and intense heat. Other typical symptoms of ED that should be treated symptomatically include xerostomia, atopic dermatitis, and dryness of the nose and eyes. It is recommended that all hypo hidrotic ED patients see a dentist. Hypo hidrotic ED patients are more prone to melancholy, low self-esteem, and insecurity due to their unusual physical characteristics and lack of social acceptance.⁴

Managing Hypohidrotic Ectodermal Dysplasia involves a multidisciplinary team approach, patient education, and early intervention in selected patients. All patients with this condition, and their families, should be offered genetic counselling and testing.⁵

4 Conclusion

Classification of EDs is difficult due to their heterogeneous grouping of inherited disorders with overlapping symptoms. Those who experience clinical indications face significant social challenges. It is typically necessary to diagnose early and formulate rehabilitation through interdisciplinary teamwork. A multidisciplinary team of physicians, comprising pediatricians, ENT specialists, and skilled dentists, is frequently required to treat the patients. Furthermore, to help ED sufferers maintain their sense of self-worth, psychologists' assistance is typically necessary.

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