HYPOHIDROTIC ECTODERMAL DYSPLASIA - A CASE STUDY

Geetha Varghese[1] , Pradeesh Sathyan[2]

[1] *Professor,* [2] *Sr. Lecturer,*
Dept.of Oral Pathology, Sree Mookambika Institute of Dental Sciences,Kulasekharam, Tamil Nadu
Corresponding Author : Dr. Geetha Varghese, 'Kattunilathu', Vattiyoorkavu, Trivandrum – 13
Email id: tvm.provide@gmail.com

Abstract

Hereditary Ectodermal Dysplasia is an inherited disorder involving skin, teeth, hair and nails. Hypohidrotic ectodermal dysplasia or Anhydrotic ectodermal dysplasia is the most common syndrome among this large group of heriditory disorders. Hypohidrosis, hypotrichosis and hypodontia constitute the main symptoms of the syndrome. The case study of a 17 year old boy, a vibrant youngster with no positive family history is presented. The pattern of genetic inheritance is also discussed.

Introduction

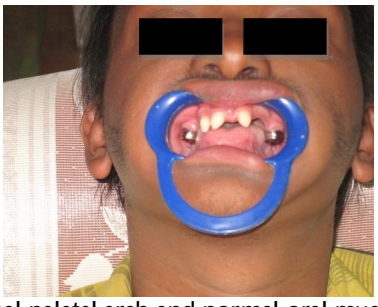
Hereditary ectodermal dysplasia is characterized by defective formation of one or more structures derived from ectoderm (1). It was first described by Thurnam in 1848 and was coined by Weech (2) in 1929. Freire-Maia and Pinheiro described numerous varieties of ectodermal dysplasia involving all possible Mendelian modes of **inheritance** (3)**.** Of these Hypohidrotic ectodermal dysplasia is the most common and estimated to affect atleast one in 17000 people worldwide. Before birth, these disorders result in the abnormal development of structures including skin,hair ,nails ,teeth and sweat glands.

Case study

A seventeen year old 12th grade male student in Trivandrum born of non consanguineous marriage presented with congenital absence of teeth. Dental examination revealed complete absence of mandibular teeth and partial anodontia in the maxillary arch. The maxillary anteriors (13, 11, and 21) were conical in shape. Full metal crowns were seen on the maxillary molars (16, 26).
Dental history revealed delayed eruption and partial anodontia in primary dentition as well. Clinical examination of the patient showed very fine and brittle hair on the scalp, large and oblique set ears as well as complete absence of eyebrows and even eyelashes.

Frontal bossing, slightly saddled nose and full and everted lips were also noted. The patient also gave a clinical history of almost complete absence of sweating from birth.

The combined dental and clinical findings pointed towards a diagnosis of hypohydrotic ectodermal dysplasia. No positive family history was found. He is the second child and the only affected member in the family. He is having

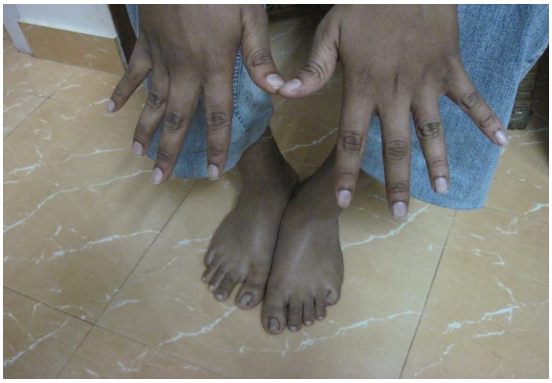


normal palatal arch and normal oral mucosa. His nails are unaffected.

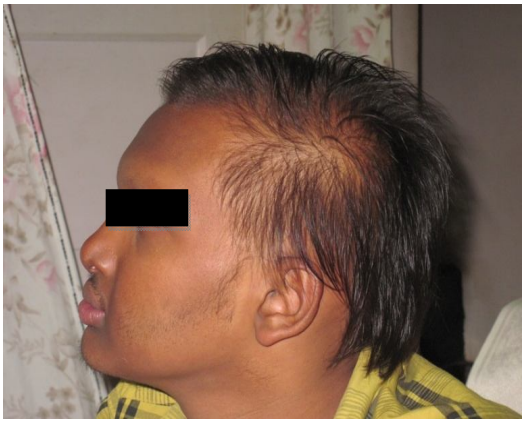
The patient had a history of removable partial denture use, but the inconvenience caused by the constant need for refabrication of dentures with the growth of his alveolar ridge

prompted him to discard them completely and seek more permanent dental options. After undergoing an alveolar ridge appraisal, the patient is waiting for implant prosthesis.

Discussion:



The ectodermal dysplasias (EDs) are congenital, diffuse and non progressive disorders. More than 192 distinct disorders have been described till date (4). It is



typically inherited as a cross-linked recessive trait so that the frequency and severity of the condition is more pronounced in males than in females. It was redefined by Freire- Maia as a developmental defect which at embryonic level affects the ectoderm and therefore the tissues and structures derived from it. Thus it affects the development of keratinocytes and cause aberrations in the hair, sebaceous glands, ecrine and apocrine glands, nails, teeth, lenses and conjunctiva of the eyes, anterior pituitary gland, nipples and the ears(5). From the **clinical** point of view two main forms have been distinguished:

1. Hypohidrotic form/Christ-Seimens-Tourian Syndrome.
2. Hidrotic form/Clouston syndrome.

The Hypohidrotic form exhibits the classic triad-hypohidrosis, hypotrichosis and hypodontia. Usually X-linked recessive inheritance is seen. Males are affected severely, while females show only minor defects. In the hidrotic form teeth, hair and nails are affected. The sweat glands are usually spared. It is usually inherited as an autosomal dominant trait. Other inheritance modalities like autosomal recessive have also been reported(1,6).

TABLE 1 - Differences between the hydrotic and hypohydrotic forms of ectodermal dysplasia(7)

	Hydrotic	**Hypohydrotic**
Mode of Inheritance	Most often autosomal dominant	Most often autosomal recessive
Scalp Hair	Soft, dawny, color is darker	Fine in texture, fair and short
Teeth	Anodontia to hypodontia	Anodontia to hypodontia
Lips	No abnormality	Protruding
Sweat glands	Active	Reduced to absent
Nasal bridge	No flattening	Underdeveloped
Nails	Dystrophic nails	No abnormality
Eyebrows	Frequently absent	Absent
Eyelashes/ Pubic/ Axillary hairs	Scanty/absent	Variably affected

In the hypohidrotic form.the skin is soft, thin and dry. Partial or complete absence of sweat glands is responsible for the inability to perspire and accounts for intolerance to heat and frequent hyperthermia. The sebaceous glands are also defective or absent (8, 10).Palms and soles are hyperkeratotic; psuedorrhagades are present around the eyes. Atopic eczema is common, especially in flexures during early childhood. In a new born, a "plastic wrap" appearance of the skin is characteristic of hypohydrotic type. In the hydrotic type, sweat glands develop partially; ducts may be formed but secretory coils are absent and there are reduced number of sweat pores[1].Scalp hair is often fine, stiff and short. Eyelashes and eyebrows are scanty and often missing (8, 11).

The characteristic facial features are: frontal bossing, depressed nasal bridge, prominent supra orbital ridges, prominent and obliquely set ears , midface is depressed, the lower third of the face appears small due to lack of alveolar bone development, lips are protuberant(8,12). A cephalometric study by Vierucci and co-workers have shown significant differences in craniofacial features of unaffected and affected children (13). Children with hypohidrotic type ectodermal dysplasia showed maxillary retrusion due to sagitally under developed maxilla, forward and upward displacement of the mandible and collapsed lower anterior facial height.

In the oral cavity, the most striking feature is oligodontia. The teeth that are present have abnormal crown form. Teeth in the anterior region of maxilla and mandible are conical in shape (9, 10). There is a wide midline diastema and hypoplastic labial frenum. Cuspids are usually normal. Commonly, there is only one molar tooth in the second molar region which usually exhibits a bud crown form. Consistent variations in the number and crown forms of teeth occur which appear to be a characteristic dental phenotype for ectodermal dysplasias with different modes of inheritance. This is demonstrated especially in autosomal recessive condition where there is a total absence of permanent teeth with or without taurodontism of primary molars(1). Cases have been reported where both primary and permanent dentition were congenitally missing (11, 12, 13).

Genetic studies of more than 300 cases have revealed X linked mode of inheritance with its gene locus being Xq11-21.1, the gene is carried by the female but manifested in the male. However there are reports of multiple siblings being affected and of females suffering with this condition (5). Mutations in EDA,EDAR and EDARADD genes are now identified to cause Hypohidrotic ectodermal dysplasia. These genes provide instuctions for making proteins that work together during embryonic development. These proteins form part of a signalling pathway that is critical for the interaction between the two germ layers the ectoderm and mesoderm. In the early embryo these cell layers form the basis for many of the body's organs and tissues. Ectoderm-mesoderm interaction is essential for the formation of several structures that arise from the ectoderm including skin, hair, nails, teeth and sweat glands. Mutation in the EDA,EDAR or EDARADD gene prevent the normal interaction between ectoderm and mesoderm and impair the normal development of hair, sweat glands and teeth. The improper formation of these ectodermal structures leads to the characteristic features of Hypohidrotic ectodermal dysplasia(16,17).

The presentation of thin eyebrows and partial anodontia observed in our case is in agreement with existing literature (14). Whereas, the observation of normal form and shape of finger and toenails in the present cases is in accordance with previous observation of Shaw. The proband was the only child of his parents exhibiting fetures of ED. No similar case of ectodermal dysplasia has been identified among the relatives, which suggests that the propositus was probably a fresh mutation or due to translocation of genes as was suggested in a few other literature(7).

Conclusion:

In developed countries diagnosis pertains to laboratory identification of genes

(15) and mode of inheritance of mutant genes associated with recessively X chromosome or autosomal dominant or recessive. This may be difficult in developing countries like India where such facilities are insufficient and it requires further probing through genetic analysis. It is to be noted that absence of a positive family history should not be a factor in causing any diagnostic dilemmas with respect to ectodermal dysplasia, a condition that shows multiple modes of inheritance.

References:

1. Rajendran R., Sivapathasundaram B. Shafer's textbook of oral pathology. 5th ed. Philadelphia: Saunders; 1983
2. Weech AA. Hereditary ectodermal dysplasia. Amer J Dis Child. 1929;37 (6):766.
3. Hall K.R. Pediatric orofacial medicine and pathology; 4th ed; London; Chapman and Hall; 1994:163.
4. Shah KN, Mckinster CD. Ectodermal Dysplasia. eMedicine Article last updated: Nov 8, 2006
5. Itthagarun A, King NM. Ectodermal Dysplasia: a review and case report. QuintessenceInt. 1997 Sep;28(9):595-602.
6. Lowry R.B., Robinson G.C., Miller J.R. Hereditary ectodermal dysplasia: symptoms, inheritance patterns, differential diagnosis, management Clin.Pediatr 1966;5:395-402.
7. Jyothi Sharma, GP Mamatha. Hereditary ectodermal dysplasia:diagnostic dilemmasRev Clín Pesq Odontol. 2008 jan/abr;4(1):35-40
8. Suprabha B.S Hereditary ectodermal dysplasia : a case report J Indian Soc Pedo Prev Dent March 2002
9. Sushma N Ramraje*, Minal Wasnik**, Yasmin A Momin Anhidrotic Ectodermal Dysplasia- A Report of Two Cases Bombay Hospital Journal, Vol. 51, No. 2, 2009
10. Crawford P.J.. Aldred M.J., Clarke A Clinical and radiographic dental findings in x-linked hypohidrotic ectodermal dysplasia J.Med.Gen 1991;28:181-185.
11. Franchi L, Branch! R., Tollaro I. Craniofacial changes following early prosthetic treatment in a case of hypohidrotic ectodermal dysplasia with complete anodontia J.Dent.Child 1998;65:116-121.
12. Shaw R.M. Prosthetic management of hypohidrotic ectodermal dysplasia with anodontia: Case report Aust.Dent J 1990; 35: 113-116.
13. Vierucci S., Baccetti T., Tollaw I. Dental and craniofacial findings in hypohidrotic ectodermal dysplasia during primary dentition phase J. Clinical Ped Dent 1994; 18: 291-297.
14. El- Tony MK, Fetelh RM. Hereditary hypohidrotic ectodermal dysplasia with anodontia: a case report. Saudi Dent J. 1994;6(1):31-34
15. Kumar A, Eby MT, Sinha S. Jasmin A, Chaudhary PM. The ectodermal dysplasia receptor activates the nuclear factor-kappa b JNK and cell death pathways and binds to ectodysplasin. A J Biol Chem. 2001 Jan 26;276(4):2668-77. Epub 2000 Oct 16.
16. DrogenullerC,DistIO,LeebT X-linked anhydrotic ectodermal dysplasia in men,mice and cattle Genet Sel Evol2003
17. Gaide O. New development in the history of Hypohidrotic ectodermal dysplasia. Dermatology 2003,207(2)123-4

Source of Support: Nil, **Conflict of Interest**: None declared