A Case Report

Ectodermal Dysplasia: A Case Report & Literature Update

Divya Rai[1], Padmanidhi Agarwal[2], Shally Mahajan[3]

Senior Resident[1] Department of Dentistry, Dr.RMLIMS, Lucknow
Assistant Professor[2] Dr RMLIMS, Lucknow
Professor[3] Department of Dentistry, Dr. RMLIMS

Abstract

Ectodermal dysplasia is a rare hereditary disorder characterized by abnormal development of certain tissues and structures of ectodermal origin. This condition is of importance to dentists as it affects the teeth resulting in hypodontia or anodontia. It can also affect the skin, the lens or retina of the eye, parts of the inner ear, the development of fingers and toes, the nerves and other parts of the body. Apart from having difficulties in eating and speaking, young affected individuals can also feel that they look different from their peers, resulting in low self-esteem.The aim of this paper is to report one such case and discuss the etiology, genetis, clinical manifestations and treatment options of this hereditary disorder.

Keywords- Ectodermal dysplasia, syndrome, anomaly, hypodontia, anodontia

Access this article online

Website: www.healtalk.in

DOI: 10.5281/zenodo.7408021

Quick Response Code:

INTRODUCTION

The term ectodermal dysplasia (ED) is used to designate a heterogenous group of disorders characterized by a constellation of findings involving a primary defect of the skin, teeth and appendageal structures including hair, nail, exocrine and sebaceous glands.[1]

Hereditary ED is characterized by defective formation of one or more structures derived from ectoderm. [(2)] It is characterized by the triad of signs including sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hyperhidrosis).[(3)]

We hereby describe a rare case of ED in a 5-year-oldwith typical features of the syndrome.

CASE REPORT

A 5-year-old female patient came to the Out Patient Department of the Department of Dentistry, Dr. Ram Manohar LohiaInstitute of Medical Sciences, Lucknow, with the chief complaint of multiple missing teeth in relation to upper and lower alveolar arches. Patient's mother reported no history of trauma or infection in the past. The lack of primary and permanent teeth in the oral cavity were causing dietary and speech problems for the patient.

They reported no family history and no evidence of similar problem either in the maternal or paternal side of the patient's family. Both the children had achieved developmental milestones in time, were vaccinated as per the advised schedule and had normal intelligence.No visual or auditory problems were evident.

General Examination revealed nail dystrophy and dry skin. soft, dry and light-coloured skin on the face and on the upper and lower extremities, sparse hair on the scalp, eye brows and eye lashes.Her extra oral examination revealed her to be having frontal bossing, thick and protuberant dry lips and a saddle nose. (Figure 1a,b)

Upon intraoral examination, 7 deciduous teeth were present(including right upper incisors, right upper first molar, right upper first molar, left lower central incisors, left lower lateral incisors, left lower first molar, and right lower central incisors), and she also had a cone-shaped teeth. Some of the teeth were fused. (Figure2)

Orthopantomogram (OPG) was taken which revealed multiple missing teeth, generalized interdental bone loss, and developing tooth buds. (Figure 3) Hand wrist radiograph revealed shortened middle and distal phalanges of all fingers.

How to cite this article: Rai et al.: Ectodermal Dysplasia: A Case Report & Literature Update, HTAJOCD 2022;Nov-Dec (2):18-21.

On the basis of clinical and radiographic features, the final diagnosis of ED was achieved.

Dental rehabilitation techniques in terms of prosthetic options once the patient is a little older were explained to the patient's mother. Counselling was done regarding the disorder to alleviate any anxiety in the patient or her family and her psychiatric reference was also sought. She has been advised regular 6 monthly follow up in both psychiatry and dental departments.

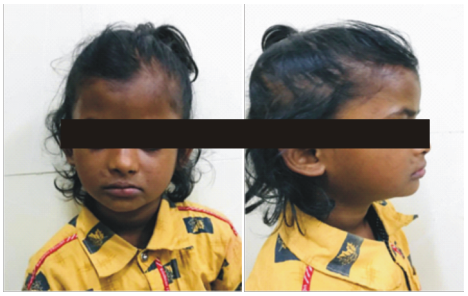

Figure 1: Extraoral picture showing sparse hair, saddle nose & in (a) frontal and (b)Lateral profile

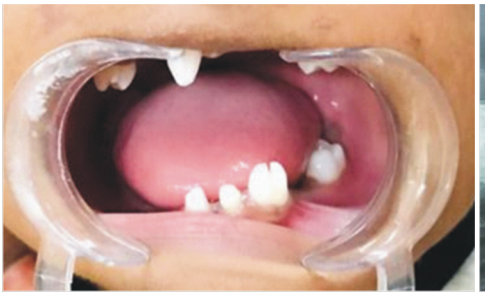

Figure 2: Hypodontia & fusion evident on intraoral examination

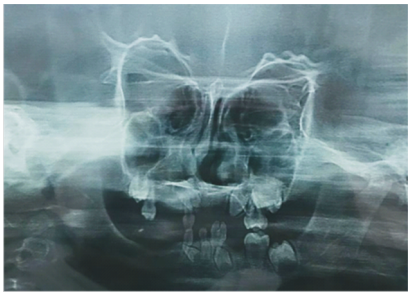

Figure 3: OPG showing hypodontia, fusion and missing tooth buds of permanent teeth

DISCUSSION

ED was first described by Thurnam in 1848[4] and coined by Weech in 1929.[5] These disorders result in the abnormal development of structures including skin,hair,nails, teeth and sweat glands. The incidence of this condition is 1:100,000 in males and the carrier's incidence is around 17.3 in 100,000 women.[6]

Broadly, ED is classified into either group A disorders, which were manifested by defects in at least 2 of the 4 classic ectodermal structures and group B disorders which were manifested by a defect in one classic ectodermal structure in combination with a defect in one other ectodermal structure (i.e. ears, lips).[7]Depending on the number and functionality of the sweat glands, two major types of ectodermal dysplasia have been classified - X-linked anhidrotic or hypohidrotic(HAED) type (Christ-Siemens-Touraine syndrome)and Hidrotic(HED) type (Clouston's syndrome).[5] of these, ChristSiemens-Touraine syndrome is the most frequently reported manifestation of ectodermal dysplasia.[(8)] of these Hypohidrotic ED is the most common and estimated to affect at least 1 in 17000 people worldwide.[9]

Different types of ectodermal dysplasia are caused by the mutation or deletion of certain genes located on different chromosomes. Because ectodermal dysplasia is caused by a genetic defect it may be inherited or passed down the family line. In some cases, they can occur in people without a family history of the condition, in which case a de novo mutation would have occurred. Mutations in the EDA, EDAR and EDARADD genes are known to cause HED. EDA is the only gene known to be associated with X-linked HED (XLHED). 95% individuals with HED have the X-linked form. The genes EDAR and EDARADD are known to be associated with both autosomal dominant and autosomal recessive forms of HED. Mutations in these genes account for 5% of HED [(10).]These genes are responsible for the formation of several substrates required for the activation of the tumour necrosis factor α-related signalling pathway, the WNT-signalling pathway, and the nuclear factor-κB pathway, involved in ectoderm-mesoderm interactions, differentiation of ectodermal appendages and organogenesis during the initiation of embryonic development.[(11)]

Typical facial features include frontal bossing, sunken cheeks, a saddle nose, thick and everted lips, wrinkled and hyperpigmented skin around the eyes, and large low-set ears. Dental manifestations include conical or pegged teeth, hypodontia or complete anodontia, and delayed eruption of permanent teeth. Eccrine sweat glands may be absent or sparse and rudimentary, particularly in those with hypohydrotic ED. In some cases, mucous glands are absent in the upper respiratory tract and in the bronchi, esophagus, and duodenum.[(12)] Other common signs are short stature, eye abnormalities, decreased tearing and photophobia.[(3)]Other common manifestations such as atopic dermatitis, xerostomia, dryness of eyes and nose should be treated symptomatically.[(13)] Oral traits of ectodermal dysplasia may be expressed as anodontia or hypodontia, with or without a cleft lip and palate.[(16)]

Clinically hypohidrotic/anhidrotic ED is characterized by hypotrichosis, hypo/adontia, dyshidrosis (abnormal sweating), and facial dimorphism.[(14)] On the other hand, hidrotic ED is characterized by the triad of onychodysplasia, hypotrichosis, and palmoplantar hyperkeratosis.[(14)] The clinical presentation of our patient was consistent with a case of hypohidrotic ED.

Mortality is as high as 30% in the first 3 years of life in children with hypohidrotic ED, due to numerous complications such as failure to thrive, pulmonary infections, and hyperthermia. Hence, additional care must be provided to infants and young children by the treating physician. After 3 years of life, life expectancy is normal.[(15)]

In the present case scenario, the maxillary deciduous right and left central incisors are replaced by the existing permanent incisors. The literature documents that there are generally more teeth in the maxilla than the mandible, although both thejaws can be toothless .[(17)] The present case series clearly depicts the presence of maxillary teeth and complete absence of mandibular teeth. Besides the delay in teething, the teeth erupted appear to be conical or peg shaped in appearance with upper incisors and cuspids affected more commonly [(3).] The presence of conical shaped maxillary incisors in the siblings further strengthens the diagnosis of Ectodermal Dysplasia.Interestingly, in this case the growth of the jaws was not affected, however resulted in underdeveloped alveolar

ridges as witnessed in radiographic examination(OPG). This is attributed to the fact that in the absence of teeth the alveolar process does not develop much resulting in reduction of normal vertical dimension.

Phenotypic tests such as assessment of sweating and dental findings aids to identify possible female carriers. Two methods of assessment of sweating have been developed: The first sweat test is performed on the backs of the carrier female and gives a V- shaped pattern of streaks that refers to the lines of Blaschk. [18] The other method is to make counts of the sweat pores along ridges of the finger tips or palms. Sweat pore counts using yellow starch iodine, pilocarpine iontophoresis may document hypotrichosis.[18]

Combination of both dental examination and sweat testing enhances clearly the chances of making a correct diagnosis of identifying female carriers. Dental radiographs can provide useful additional information and can be a simple screening test for carrier states. Differential diagnosis of alopeciaareata, focal dermal hypoplasia,incontinentiapigmenti, and dyskeratosis congenita should be considered[19]

So, based on all the above possible clinical and radiological findings found inpatient, a diagnosis of X-linked hypohidrotic ectodermal dysplasia had been achieved.

Dental referral is warranted in all patients of hypohidrotic ED. In childhood, dentures are the primary line of treatment for the dental abnormalities. These dentures are regularly evaluated by the paediatric dentist as per the child's growth and development and are modified/replaced accordingly. In older individuals, dentures, dental implants, and orthodontia are usually the preferred treatment options.

Patients with hypohidrotic ED may suffer from low self-esteem, insecurity, and depression due to their unusual physical appearances and lack of social acceptance.([10,18] Hence, psychological counselling should be advised on a regular basis. Use of wigs in patients with severe alopecia, and early initiation of dental prosthetics may improve their cosmetic appearance. Consultation with a speech therapist and an otolaryngologist is warranted if abnormities in phonetics and word-articulation are detected.

Recent literature highlights that intravenous injection of recombinant EDA-A1 to newborn dogs with X-linked hypohidrotic ED has found to restore the growth of their teeth, skin structures, and mucous glands. Furthermore, intra-amniotic injections of recombinant EDA-A1 in pregnant mice partially improved the phenotype of the X-linked hypohidrotic ED newborn mice.[20] This recombinant EDA-A1 at present is in Phase-II clinical trials and is being administered to newborn males with hypohidrotic ED to hopefully alleviate some of their symptoms. Further studies are warranted in this regard.

Limitationsof this case included the inability to carry out any gene testing for the patient due to financial constraints.

CONCLUSION

EDs are rare genetic disorders that have many overlapping features and it is difficult to classify them. This case report highlights the need for adequate knowledge about this disease amongst physicians and dentists for its diagnosis. The clinical manifestations of ED cause significant oral and general body functions as well as social problems in affected individuals. The success of management lies inearly diagnosis and prosthetic rehabilitation by the multidisciplinary approach. The role of dentists is very important for management and psychologist support is mandatory for behavioural management of the child.Further studies are ongoing to diagnose and treat the syndrome in vitro.

Declaration of Patient Consent/Ethical Statement

These authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal patient identity, but anonymity cannot be guaranteed.

Conflicts of Interest

There are no conflicts of interest.

Acknowledgment

We thank the patient and his parents for their cooperation and providing the consent for publishing the photographs.

Funding

No source of funding

REFERENCES

1. Gopinath VK, Manoj KM, Mahesh K. Hypohidrotic ectodermal dysplasia: a case report. J Indian Soc PedodPrev Dent. 1999 Sep;17(3):90–92.
2. Sharma J, MamathaGP. Hereditary Ectodermal Dysplasia :diagnostic dilemmas Rev ClínPesqOdontol. 2008 jan/abr;4(1):35-40
3. D. Ayesha Thabusum1, N. Rajesh, R. Sudhakarareddy, T. Ramesh.Ectodermal Dysplasia – A Case Study of Two Identical Sibilings International Journal of Dental Sciences and Research, 2014, Vol. 2, No. 6, 175-9.
4. Jananee J, Satish Kumar M, Balaji Sumathi. Ectodermal Dysplasia – A case report. Indian Journal of Multidisciplinary Dentistry 2012; 2(2):465-467.
5. Weech AA. Hereditary ectodermal dysplasia. Amer J Dis Child. 1929;37 (6):766.178
6. Mortier K, Wackens G. Ectodermal Dysplasia anhidrotic. OrphanetEncyclopedia. Sept 2004.
7. Marwaha M, Nanda KDS. Ectrodactyly, ectodermal dysplasia, cleft lip, and palate (EEC syndrome). Contemp Clin Dent.April2012.
8. Viera K A, Teixeira M S, Guirado C G, Gaviao M B. Prosthodontic treatment of hypohidrotic ectodermal dysplasia with complete anodontia: case report. Quintessence Int 2007; 38: 75-80.

9. Geetha Varghese1, PradeeshSathyan. Hypohydrotic Ectodermal Dysplasia - A Case Study. Oral Maxillofac Path. 2(1) Jan- Jun 2011.
10. Deshmukh S, Prashant S. Ectodermal Dysplasia: A Genetic Review international Journal of Clinical Pediatric Dentistry.5(3):September-December 2012;197-202.
11. Trzeciak WH, Koczorowski R. Molecular basis of hypohidrotic ectodermal dysplasia: An update J Appl Genet. 2016; 57:51–61.
12. Siew-Yin CheeHsing Wanga,1,2 Wei-De Lina,1,3 and Fuu-Jen Tsai. Ectodermal dysplasia (ED) syndrome- 2014 Nov 26.
13. Joseph S, Cherackal GJ, Jacob J, Varghese AK. Multidisciplinary management of hypohydrotic ectodermal dysplasia – A case report Clin Case Rep. 2015; 3:280–6
14. Meshram GG, Kaur N, Hura KS. A case report of hypohidrotic ectodermal dysplasia: A mini-review with latest updates. J Family Med Prim Care 2018 Jan-Feb; 7(1): 264–266.
15. Blüschke G, Nüsken KD, Schneider H. Prevalence and prevention of severe complications of hypohidrotic ectodermal dysplasia in infancy. Early Hum Dev. 2010;86:397–9.
16. Ectodermal Dysplasia with Anodontia: A Report of Two Cases Mehmet Bani,a Ali MelihTezkirecioglu,bNeseAkal,c and Tamer Tuzunerd 2010 EUR J DENT.Apr; 4(2): 215–222.
17. Aswegan A.L., Josephson K.D., Mowbray R., Pauli R.M., Spritz R.A., Williams M.S.: Autosomal dominant hypohidrotic ectodermal dysplasia in a large family. Am. J. Med. Genet. 1997; 72: 462-467
18. Clarke A., Burn J: Sweat testing to identify female carriers of Xlinkedhypohidrotic ectodermal dysplasia. J Med Gen 1991(28): 330-333
19. Prerna Taneja Mathur, Rahul Paul, Priyank Banthia, TapasayaJuneja Kapoor Understanding Ectodermal Dysplaisa. Indian Journal of Dental Sciences 2013 Supplementary Issue, Issue:4, Vol.: 5
20. Hermes K, Schneider P, Krieg P, Dang A, Huttner K, Schneider H, et al. Prenatal therapy in developmental disorders: Drug targeting via intra-amniotic injection to treat X-linked hypohidrotic ectodermal dysplasia. J Invest Dermatol 2014;134:2985-7.