



## ECTODERMAL DYSPLASIA- AN INCIDENTAL DIAGNOSIS AT AN UNUSUAL AGE


**K. Saraswathi Gopal, A. Vani Anusha**

<sup>1</sup>Professor and Head of the Department of Oral Medicine and Radiology, <sup>2</sup>Department of Oral Medicine and Radiology, Meenakshi Ammal Dental College and Hospital, Chennai - 600 095, Tamilnadu, India.

### ABSTRACT

Ectodermal dysplasia is a hereditary disorder that occurs as a consequence of disturbances in the ectoderm of the developing embryo. The triad of nail dystrophy, alopecia or hypotrichosis and palmoplantar hyperkeratosis is usually accompanied by a lack of sweat glands and a partial or complete absence of primary and/or permanent dentition. A case report illustrating the features of hypodontia, hypohidrosis and partial hypotrichosis is presented, the diagnosis being incidental at an unusual age.

**Key words:** Ectodermal dysplasia, Hereditary, Hypodontia, Hypohidrosis, Hypotrichosis, Christ-Siemens- Touraine syndrome.

| Access this article online  |   |                   |
|---|---|-------------------|
| Home page:<br><a href="http://www.mcmed.us/journal/ijacr">http://www.mcmed.us/journal/ijacr</a>               | Quick Response code<br> |                   |
| DOI:<br><a href="http://dx.doi.org/10.21276/ijacr.2018.5.1.1">http://dx.doi.org/10.21276/ijacr.2018.5.1.1</a> |   |                   |
| Received:05.01.18   | Revised:22.01.18  | Accepted:13.02.18 |

### INTRODUCTION

Ectodermal dysplasia, as first described by Thurman [1] is a hereditary disorder occurring as a consequence of disturbances in the ectoderm of the developing embryo. The triad of nail dystrophy (onchodysplasia) [2], alopecia or hypotrichosis (scanty, fine light hair on the scalp and eyebrows), and palmoplantar hyperkeratosis is usually accompanied by a lack of sweat glands (hypohidrosis) and a partial or complete absence of primary and/or permanent dentition. Ectodermal dysplasia represents a large and complex group of diseases comprising more than 170 different clinical conditions[3]. The incidence of this condition is 1:100,000, with a mortality rate of 28% in males up to 3 years of age[4-6]. When at least 2 types of abnormal ectodermal

features occur such as malformed teeth and extremely sparse hair, the patient is diagnosed with ectodermal dysplasia syndrome. The following is a case report presenting with a partial expression of hypohidrotic ectodermal dysplasia which was an incidental diagnosis at an unusual age.

### CASE REPORT

A 42 year old male patient reported to the department of oral medicine with a chief complaint of pain in the left lower back tooth region for the past one week. Intra oral examination revealed dental caries in 37 that was tender on percussion, edentulous area in relation to 11,12,21,22 and 31,32,41,42 region. Patient history revealed congenital absence of the maxillary and mandibular incisors both in deciduous and permanent dentition. Patient revealed that none of the family members had similar and associated features. On general examination and review of systems, no apparent systemic illness was noted and all the vitals were within the normal limits. Extra oral features of saddle nose, thinning of eyebrows were elicited. Scalp hair appears to be normal

### Corresponding Author

**K. Saraswathi Gopal**

Professor and Head of the Department of Oral Medicine and Radiology, Meenakshi Ammal Dental College and Hospital, Chennai - 600 095, Tamilnadu, India

**Email:** dr.saraswathik@yahoo.co.in

and patient presented with involutinal alopecia with frontal baldness (fig 1). The skin over flexor surfaces of hands and feet was apparently dry, scaly with scanty distribution of hair (fig.2). The patient gave a history of hyperthermia and heat intolerance on exposure to temperatures and these episodes occur seasonally. Patient was subjected to routine preliminary investigations like haematogram and chest X-ray which revealed no absolute

abnormalities. Patient presented with a panoramic radiograph which revealed edentulous areas in the 11,12,21,22 and 31,32,41,42 regions (fig.3). Considering the dermatological findings of hypohydrosis, hypotrichosis of hand and feet and intra oral findings of oligodontia, a diagnosis of partial expression of hypohydrotic ectodermal dysplasia was given.

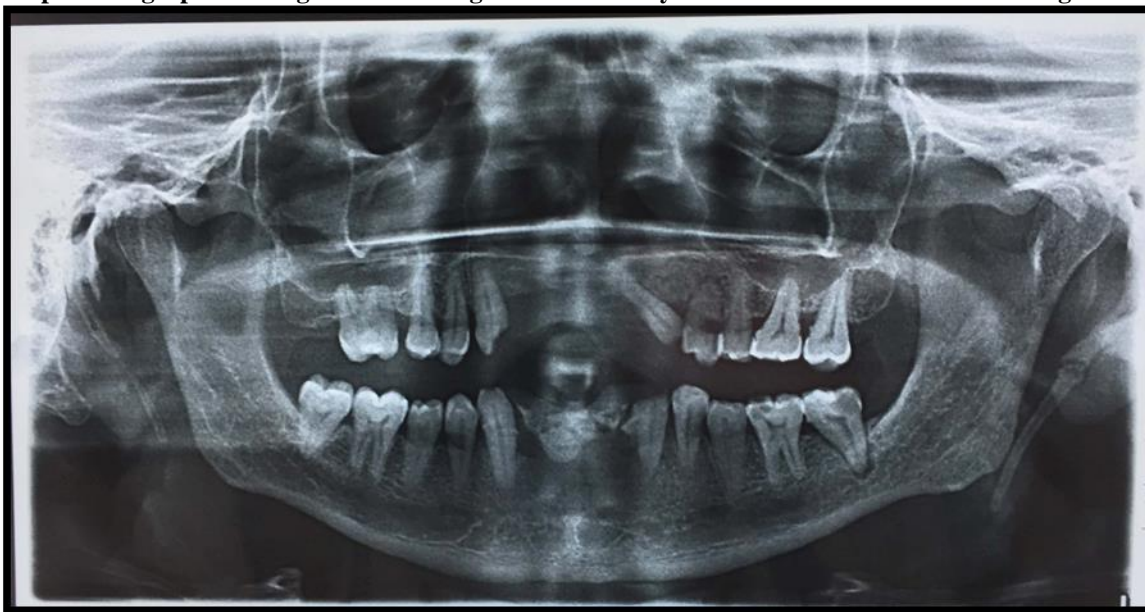
**Fig 1. Involutinal alopecia with frontal baldness**



**Fig 2. Dry scaly skin over the hands**



**Fig 3. Orthopantamograph showing edentulous regions in maxillary and mandibular anterior teeth region**



## DISCUSSION

Although first published case was reported almost as early as in 1848 by Thurnam, it was in the year 1929 that the term 'ectodermal dysplasia' was coined by Weech[1]. The incidence of Ectodermal Dysplasia is approximated to be 0.7 to 1/100,000 births [1, 2]. Freire-

Maia and Pinheiro (1973) provided an extensive review and thus proposed a provisional classification based on which ectodermal derivatives are affected [3,4]. According to their classification, '1' indicates hair dysplasia, '2' dental dysplasia, '3' nail dysplasia and '4' sweat gland dysplasia. Recently, many other classifications have been

provided. In 2001 Priolo and Laganà reclassified the Ectodermal Dysplasias into two main functional groups: (1) defects in developmental regulation/epithelial-mesenchymal interaction and (2) defects in cytoskeleton maintenance and cell stability [5]. In 2003, Lamartine reclassified the Ectodermal Dysplasias into four functional groups based on the underlying pathophysiological defect [6]. Reports relating to the EEC (ectrodactyly ectodermal dysplasia syndrome cleft lip/palate) syndrome and similar conditions show uncertainty about the mode of inheritance [7]. Autosomal dominant inheritance with variable expressivity and incomplete penetrance, as well as autosomal recessive inheritance, have been reported [8].

The various genetic forms of Ectodermal Dysplasia can be X-linked recessive, autosomal recessive or autosomal dominant yet in the present case the patient had not acquainted any admissible family history and that no akin clinical features were noticed. The aetiology is related to either mutation or deletion of certain genes located on different chromosomes. The most commonly implicated genes are EDA (ectodysplasin), EDAR (ectodysplasin receptor) and EDARADD (ectodysplasin receptor associated death domain) [1, 8]. Since in our case the patient was not subjected to genetic analysis, we could not point out the exact gene. Dental findings in ectodermal dysplasia may range from hypodontia to anodontia of the primary or permanent teeth. The congenital absence of primary teeth is relatively rare [6,9]. Nevertheless, oligodontia involving primary and permanent dentitions was elicited in the present case.

Oral rehabilitation of the ectodermal dysplasia patient is necessary to improve both the sagittal and vertical skeletal relationship during craniofacial growth and development as well as to provide improvements in esthetics, speech, and masticatory efficiency [4-9]. Although removable prostheses are the most common treatment method, dental implants are also considered to be a treatment option. In the present case, the patient was a denture user since his childhood and was totally aware of the absence of anterior teeth. No apparent facial growth

disturbances were elicited. There are 2 major types of this condition depending on the number and functionality of the sweat glands: (1) X-linked anhidrotic or hypohidrotic, where sweat glands are either absent or significantly reduced in number (Christ-Siemens- Touraine syndrome), and (2) hidrotic, where sweat glands are normal and the condition is inherited as autosomal dominant (Clouston's syndrome) [10, 11]. Since, in the present case the patient gives a history of heat intolerance for temperatures episodically which is experienced by the patient seasonally, a partial involvement of sweat glands and hypohydrosis was considered. The dentition and hair are affected similarly in both types, but the hereditary patterns, nail and sweat gland manifestations tend to differ [12, 13].

## CONCLUSION

The clinical manifestations of ectodermal dysplasia are variable. Henceforth, through history, evaluation and analysis of familial and syndromic correlations is a mandatory during the diagnosis of congenital anodontia and similar conditions which aids in appropriate treatment planning and decides the choice of management modality. Genetic counselling serves to be a preventive measure depending on the severity of expression of the syndrome.

## STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

## ACKNOWLEDGEMENT

Nil

## CONFLICT OF INTEREST

No interest

## REFERENCES

1. Gupta M, et al. (2014). Christ Siemen Syndrome. *BMJ Case Rep*, 10, 1136.
2. Deshmukh S and Prashanth S. (2012). Ectodermal dysplasia: a genetic review. *Int J ClinPediatr Den*, 5, 197–202.
3. Freire N. (1975). Nosologic groups with special reference to ectodermal dysplasia. (In press.)
4. Koteeswaran D, et al. Christ Siemen Syndrome - A Report Of Two Cases. *Jpfa*, 25, 172-175.
5. Bixler D, Spivack J, Bennett J, Christian JC. (1971). Theectrodactylyectodermal dysplasia-clefting (EEC) syndrome. Report of two cases and review of the literature. *Clin Genet*, 3, 43-51.
6. Saraswathi G, et al. (2010). Ectodermal dysplasia- two case reports and review of literature. *International Journal of Current Research*, 9(2), 47073-47077.
7. Rosenmann A, Shapira T, Cohen MM. (2009). Ectrodactyly, ectodermal dysplasia and cleft palate (EEC syndrome). Report of a family and review of the literature. *Clin Genet*, 9, 347-53.
8. Harper JI and Trembath RC. (2004). Genetics and genodermatoses. In Burns T, Breathnach S, Cox N, Griffiths C. eds. *Rook's textbook of dermatology*. 7th edn. Blackwell Science Ltd, 12.40.
9. Ohno K and Ohmori I. (2000). Anodontia with hypohidrotic ectodermal dysplasia in a young female: a case report. *Pediatr Dent*, 22, 49–52.

10. Nunn JH, Carter NE, Gillgrass TJ, Hobson RS, Jepson NJ, Meechan JG, et al. (2003). The interdisciplinary management of hypodontia: background and role of paediatric dentistry. *Br Dent J*, 194, 245-251.
11. Tarjan I, Gabris K, Rozsa N. (2005). Early prosthetic treatment of patients with ectodermal dysplasia: a clinical report. *J Prosthet Dent*, 93, 419-424.
12. Vieira KA, Teixeira MS, Guirado CG, Gavião MB. (2007). Prosthodontic treatment of hypohidrotic ectodermal dysplasia with complete anodontia: case report. *Quintessence Int*, 38, 75-80.
13. Yavuz I, Ulku SZ, Unlu G, Kama JD, Kaya S, Adigüzel O, Kaya FA, Tümen EC, Zortuk M, Bahsi E, Arslanoğlu Z. (2008). Ectodermal dysplasia: clinical diagnosis. *Int Dent Med Disorders*, 1, 1-10.

**Cite this article:**

Saraswathi Gopal K, Vani Anusha A. Ectodermal Dysplasia- An Incidental Diagnosis at an Unusual Age. *International Journal Of Advances In Case Reports*, 5(1), 2018,1-4. DOI: <http://dx.doi.org/10.21276/ijacr.2018.5.1.1>



**Attribution-Non Commercial-No Derivatives 4.0 International**