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**Case Report**

# FAMILIAL ANHIDROTIC ECTODERMAL DYSPLASIA: A RARE CASE REPORT

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**ABSTRACT**

Anhidrotic ectodermal dysplasia (EDA) is a rare genodermatosis characterized by the triad oligoanodontia, hypotricosis, hypoanhydrosis. The classical oral features of this syndrome include anodontia or hypodontia, hypoplastic conical teeth, underdevelopment of the alveolar ridges. The objective of this report is to present two male patients with hereditary ectodermal dysplasia in same family.



## INTRODUCTION

EDA, also known as Christ-Siemens-Touraine syndrome, was first described by Thurman in 1848 and later mentioned by Darwin. However, the different aspects of this disorder, such as its genetics, inheritance patterns, and clinical chacteristics, were further described by Christ Siemens, and Touraine, hence the derivation of the name of this syndrome.[1] This article reports an unusual occurrence of Anhidrotic ectodermal dysplasia (EDA) in a family .The family history in this case clearly emphasize that in most of familial EDA the mode of inheritance is X linked recessive .This article also highlight role of dental professional in treating oral complication of HED.

### *Case Report*

A 24 year old male and his younger male sibling of 4 years of age, born of consanguineous marriage presented with chief complaint of missing tooth and sparse hair over the scalp. There was complete absence of sweating since birth. As they grew older, there was failure of teeth eruption, progressive hyper pigmentation around eyes and mouth.

Patient also gives history of loss of hair mainly over scalp region. Family members were systematically evaluated to characterize the pattern of inheritance and clinical features.

The pedigree of the last two generations of the family was constructed. Based on the pedigree, we suggest that the EDA was X-linked, manifesting in males [Figure 1]. On extra oral examination inspection shows characteristic appearance of frontal bossing, loss of one third of eyebrow in face. Low set ears and parrot beak nose and depressed nasal bridge present. Lips were protuberant due to absence of permanent teeth. Skin was soft, thin, dry with hyper pigmentation around eyes and hair line region [Figure 2a]. Intraoral examination revealed hypoplastic, conical tooth in upper arch and completely edentulous lower arch [Figure 2b]. Considering the family history the patients sibling was also reviewed .On extra oral examination of 4 year old boy showed frontal bossing, a depressed nasal bridge, protuberant lips [Figure 3a]and intraoral examination reveals hypoplastic conical teeth [Figure 3b]. In view of the above short listed positive clinical findings, both the cases were provisionally diagnosed as anhidrotic ectodermal dysplasia. Differential diagnosis considered were Aplasia cutis congenita, Naegeli-franceschetti-jadassohn syndrome, Focal dermal hypoplasia syndrome.

Orthopantomograph of 24 year old male reveals complete absence of all other permanent tooth except 14,23,27and underdeveloped maxillary and mandibular alveolar ridges [Figure 4]. Lateral cephalogram of younger male sibling shows missing of teeth, except unerupted primary molar in upper right

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and left quadrant and erupted primary incisors in the maxillary anterior region [Figure 5].

Biopsy specimens of 24 year old male obtained from the outer surface of the thigh were examined for sweat glands and skin appendages. There was reduction in the number of sweat glands, hair follicles, sebaceous glands and the epidermis was thin and flattened. Eccrine sweat glands were few and very rudimentary. Skin histopathology features suggestive of ectodermal dysplasia [Figure 6].

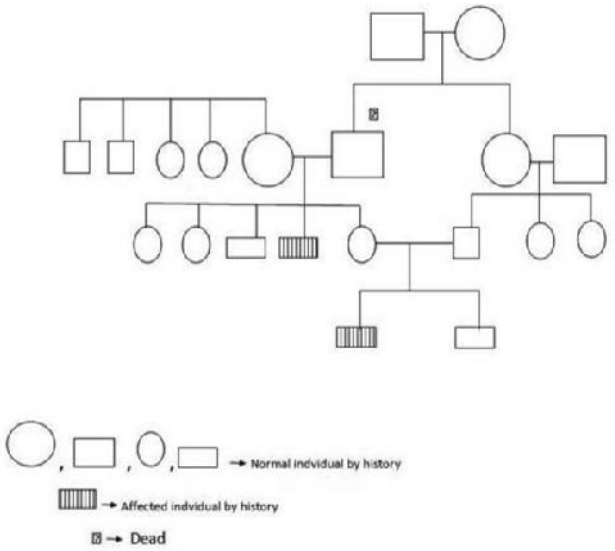


**Figure 1** Pedigree of last two generations of the family.

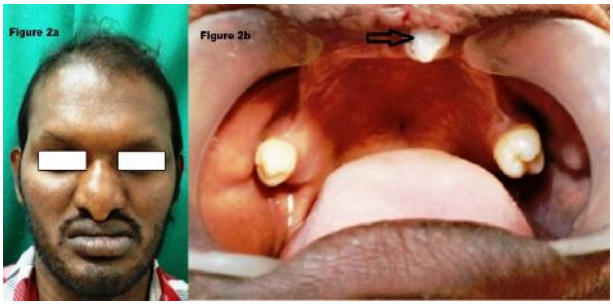


**Figure 2a** Extra oral view shows characteristic appearance of frontal bossing,low set ears and parrot beak nose and depressed nasal bridge .

**Figure 2b** Intraoral view **(Black arrow)** shows hypoplastic, conical tooth in upper arch.

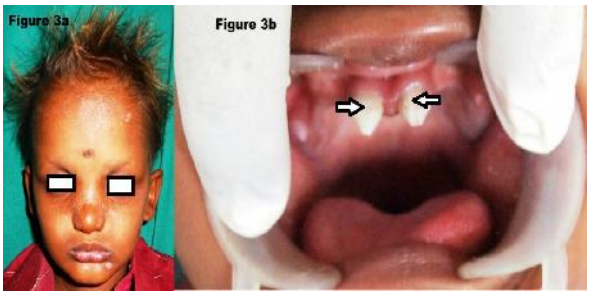


**Figure 3a** Extra oral view of 4 year old boy shows frontal bossing and depressed nasal bridge.

**Figure 3b** Intra oral view (**Black arrows)** showing hypoplastic conical teeth.

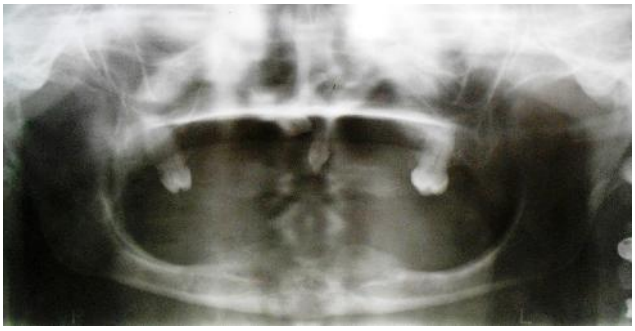

**Figure 4** Orthopantomograph shows complete absence of all other permanent tooth except 14,23,27

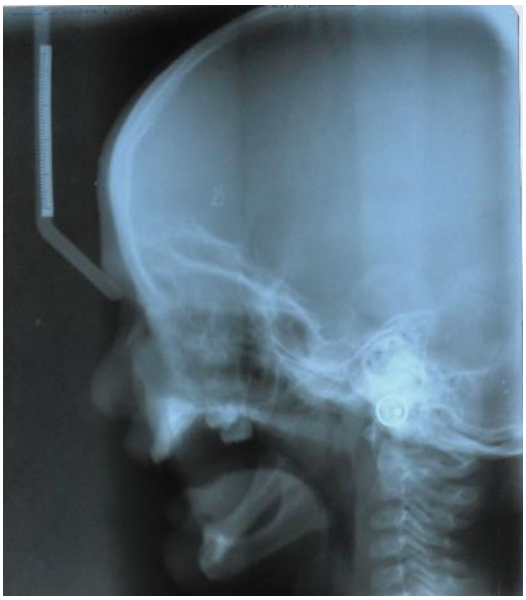

**Figure 5** Lateral cephalogram of younger male sibling shows missing of teeth, except unerupted primary molar in upper right and left quadrant and erupted primary incisors in the maxillary anterior region.

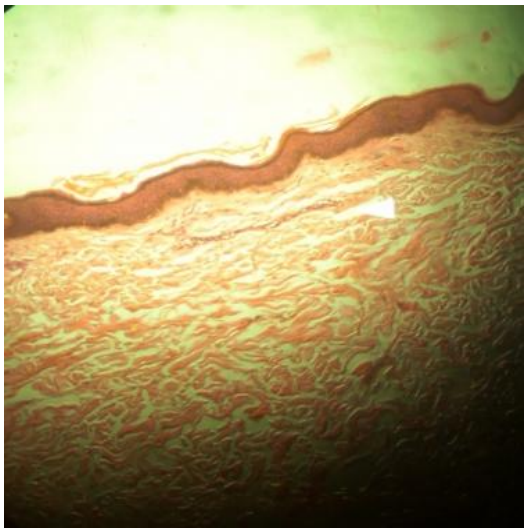

**Figure 6** Histopathological section shows a reduction in the number of sweat glands, hair follicles, sebaceous glands with thin flat epidermis.

## DISCUSSION

Ectodermal dysplasias (EDs) represent a large and complex group of diseases comprising more than 170 different clinical conditions. They are caused by impaired development of

epidermal appendages and are characterized by a primary defect in at least one of the following tissues: nails (dystrophic, hypertrophic, abnormally keratinized), hair (hypotrichosis, partial or total alopecia), teeth (abnormal or absent) and sweat glands (hypoplastic or aplastic). EDs are rare diseases with an estimated incidence of seven in 10,000 births for all EDs. They follow all possible Mendelian modes of inheritance (autosomal dominant or recessive, X-linked dominant or recessive) but sporadic cases are also described [2].

According to the state of sweat glands involvement, two major groups are distinguished:

(1) Hypohidrotic or anhydrotic (Christ-Siemens-Touriane syndrome) in which sweat glands are either absent or significantly reduced in number; (2) Hydrotic (Cloustone syndrome) in which sweat glands are normal[3]. EDA is also characterized by the triad of signs comprising sparse hair (atrichosis or hypotrichosis), abnormal or missing eeth (anodontia or hypodontia) and inability tosweat due to lack of sweat glands (anhidrosis or hypohidrosis) [4].

The oral phenotype includes multiple congenitally missing teeth, root and crown dysmorphism, mainly conical-shaped crowns, and reduced saliva flow. Microdontia is frequently observed in affected individuals. Moderate to severe taurodontism is known to preferentially affect the second primary mandibular molars in some individuals with HED [5].

Abnormalities of hair are present in all affected individuals. Most individuals have sparse, fine, slowly growing scalp hair. Some individuals are completely bald by their middle teens, whereas other individuals have normal amounts of scalp hair, though it may exhibit an abnormal texture. Sparse eyebrows and eyelashes were always found. About half of the affected individuals exhibit mild fingernail abnormalities and nail dystrophy [6].

The radiograph is of invaluable service in ascertaining the presence of hypo or anodontia and in ruling out pseudoanodontia [7].

Oral treatment of persons with HED exhibiting severe phenotypes of oligodontia or anodontia benefits increasingly from implant-supported prostheses [8]. In our cases, to improve oral function and reduce the social impairment, the patient and his sibiling were adviced for Prosthodontic rehabilitation.

## CONCLUSION

Hypohidrotic ED (HED) consist of clinical and genetic heterogenous group of disorders, characterized by the triad oligoanodontia, hypotricosis, hypoanhydrosis. Young patients with EDA need to be evaluated early by a dental professional to determine the oral complication of the condition. Genetic counseling of the involved family plays an important preventive role.

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