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HEREDITARY ECTODERMAL DYSPLASIA- WITH AN UNUSUAL AND USUAL PRESENTATION

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ABSTRACT

Hereditary ectodermal dysplasia is an X linked recessive disorder characterized by defects in the ectoderm. It is characterized by the triad of signs comprising sparse hair, abnormal or missing teeth and inability to sweat due to lack of sweat glands. But it can portray by deformity of at least two or more of the ectodermal structures, hair, teeth, nails and sweat glands. Here we present two case reports of ectodermal dysplasia having hypodontia and hypohydrosis and other classic features of this condition. Radiological manifestations of this condition is less reported which was seen in one of the case report presented as wormian bones. Case reports and review of literature are discussed.

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KEY WORDS

Ectodermal dysplasia anhydrotic (EDA), partial anodontia, sutural bones

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[I] INTRODUCTION

Hereditary ectodermal dysplasia is characterized by defective formation of one or more structures derived from ectoderm. It was first described by Thurnam in 1848 and was coined by Weech in 1929. It is remarkable that no instance has occurred of a daughter being affected [1]. Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. The X-linked recessive ED (Christ-Siemens-Touraine syndrome) is the most common disorder (80% of EDs); it affects males and is inherited through female carriers. It is characterized by the triad of signs comprising sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The lack of teeth and the special appearance were reported to be major concerns. The incidence in male is estimated at 1 in 100,000 births, the carriers-incidence is probably around 17.3 in 100,000 women [2].

A simple attempt made by Nelson included five categories, namely Hypohydrotic (anhydrotic), Hydrotic (Clouston's syndrome), EEC (Ectodactyly ectodermal dysplasia) syndrome, Rapp – Hodgkin syndrome and Robinson's disease [1].

[II] CASE REPORTS

Report-1

This patient was referred to the Department of Oral Medicine and Radiology, College of Dental Sciences, Davanagere, from a private clinic. He presented with partial anodontia. He was of 25 years and lived with this complaint since birth. He gave a history of no eruption of permanent teeth with decreased sweating and increased body temperature. His concern was regarding esthetics and loss of masticatory efficiency. There is no significant family history and medical history. On examination patient had concave facial profile due to depressed malar bone and slightly decreased lower facial height. On intraoral examination there were few retained deciduous teeth resembling canine and molars in the upper and lower arch. Based on these Hereditary Ectodermal Dysplasia was suspected. Verbal consent from the patient was taken for the photographs and radiographs. Intraoral radiographic examination revealed resorbed roots. Extraoral examination revealed sutural bones on the skull radiograph. Sutural bones were seen in the lambdoid

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region. Hand wrist and chest radiograph were normal **[Figure– 1]**.

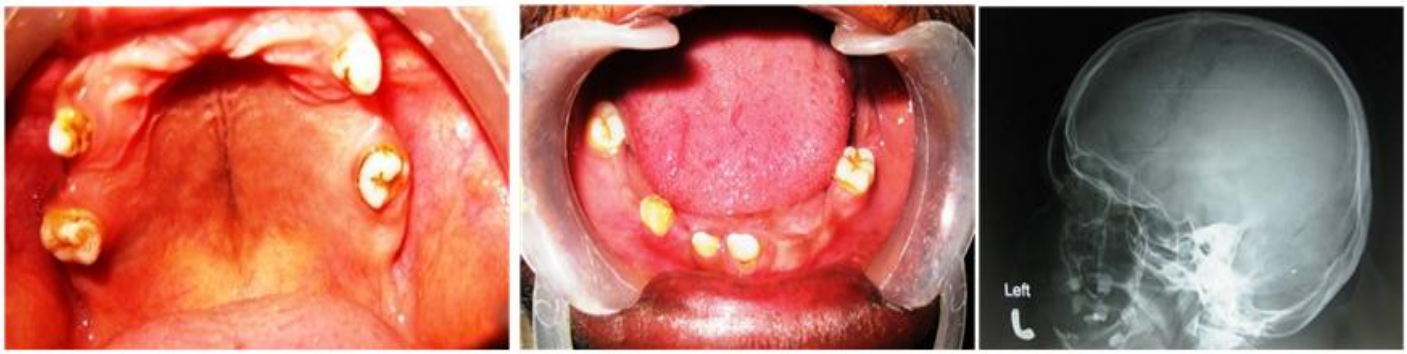

Fig: 1. Clinical photographs revealing hypodontia (several missing teeth) and radiological images depicting presence of wormian bones in the temporal region

Report-2

This patient was referred to the Department of Oral Medicine and Radiology, College of Dental Sciences, Davanagere, from a private clinic. He also presented with partial anodontia. He underwent treatment for his missing teeth but discontinued due to unknown reason and presented to our college for continuation of the treatment. There was no significant medical history. Patient gave history of similar features in the siblings. He gave a history of no eruption of few permanent teeth with decreased sweating and increased body temperature. Verbal consent from the patient was taken for the photographs and radiographs. He also presented with similar features as case 1 with unusual facies and decreased lower facial height. On intraoral examination there were missing lower incisors and upper lateral incisors **[Figure–2].**

Fig: 2. Hypodontia with crown preparation done with upper anterior teeth

[III] DISCUSSION

The prevalence of EDA is unknown; however, the incidence in male is estimated at 1 in 100,000 births although the condition is usually overlooked in infants [2].

Dental anomalies in primary dentition are frequently observed during routine dental examination, leading to orthodontic problems, including spacing or crowding of teeth, loss of arch length, deviation of the midline, increased caries risk, and esthetic problems in preschool children [3].

Hereditary hypohidrotic ectodermal dysplasia is a hereditary disease characterized by deformity of at least two or more of the

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ectodermal structures, hair, teeth, nails and sweat glands. 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 [4].

Frontal bossing, usually marked, and depressed nasal bridge give added emphasis to the small size of the face. Due to absence of teeth, and resulting reduced vertical height, the lips are protuberant. The pinnae are often outstanding [5].

A hallmark of this disorder is hypohidrosis. Inability to sweat, because eccrine sweat glands are severely reduced in number, results in intolerance to heat, with severe incapacitation and hyperpyrexia after only mild exertion [5].

The most striking oral abnormality is absence of most deciduous and permanent teeth. Maxillary central incisors and canines usually have a conical crown form and frequently one or more molars may be present these features reported were similar in the case report 1. More rarely, one or both jaws may be edentulous. Female heterozygotes exhibit reduction in numbers of teeth and smaller crown size than hemizygous males. Taurodontism is frequent which were absent in our cases. The alveolar processes do not develop in the absence of teeth [5].

Presence of sutural bones in the skull radiograph is an unusual finding in ectodermal dysplasia. Sutural bones are the intrasutural ossicles commonly seen in lambdoid, posterior sagittal, and squamosal sutures.

Usually represent normal variant, but if extensive then differential includes: PORKCHOPS6

P - Pyknodysostosis
O - Osteogenesis imperfecta
R - Rickets in healing phase
K - Kinky hair syndrome
C - Cleidocranial dysostosis
H - Hypothyroidism / Hypophosphatasia
O - Otopalatodigital syndrome
P - Primary acro-osteolysis (Hajdu-Cheney) / Pachydermoperiostosis / Progeria
S - Syndrome of Downs

The course of the treatment is to restore the function and the aesthetics of the teeth, normalize the vertical dimension and support the facial soft tissues. As long as there are no physical, psychological or social burdens, no treatment is necessary. Early placement of partial or full dentures is commonly recommended from the age of two or three years onwards. The denture must be periodically modified as alveolar growth; erupting teeth and rotational jaw growth change both the alveolar, occlusal and basal dimensions [2].

[IV] CONCLUSION

Even though there are reports concerning this condition with its classical manifestation, reports of sutural bones are minimal or lacking according to our knowledge and why this sutural bones appeared in this condition what the impact it can produce still is inconclusive. But the patient's complaint was taken into consideration and treatment was done appropriately.

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CONFLICT OF INTERESTS

Authors declare no conflict of interests.

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