Ectodermal dysplasia: report of two cases

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ABSTRACT

Ectodermal dysplasia is a hereditary disorder characterized by abnormal development of certain tissues and structures of ectodermal origin. This paper reports two cases of ectodermal dysplasia in siblings without any family history.

Keywords: Ectodermal; Dysplasia; Hypohidrotic; Siblings

Introduction

The ectodermal dysplasias are a heterogenous group of disorders with primary defect in hair, teeth, nail and sweat gland function.\(^1\)\(^2\) It is an X-linked recessive mendelian character which is rarely seen in males. Numerous types have been described and several classifications exist for this disorder. Freire-Maia defined the nosologic group of ectodermal dysplasia as any syndrome that exhibits at least two of the following features: trichodysplasia (abnormal hair), abnormal dentition, ochronodystrophosis (abnormal sweat glands).\(^3\)\(^4\) This condition is thought to occur in approximately one of 1,000 live births. The number of ectodermal dysplasia syndromes has increased to more than 170 syndromes.\(^2\)

The various syndromes associated with ectodermal dysplasia are Rapp-Hodgkin Syndrome, Strandberg- Roncheses Syndrome, Rosselli-Gulienetti Syndrome and various others. The genes responsible for this syndrome are Xq-12q 13.1, PVRL1 and GTB6.\(^5\)\(^6\) The X-linked hypohidrotic ectodermal dysplasia has full expression only in males though females outnumber the affected males; they show little or no signs of the condition.\(^3\)\(^4\) The extra oral signs may include decreased or absent sweat glands, sparse and fine hair, abnormally developed nails, prominent forehead, depressed mid face, protruberant lips, marbled pattern of skin pigmentation, atrophic rhinitis, epistaxis, saddle nose deformity, hearing loss and decreased production of body fluids.\(^7\) The commonly occurring oral manifestations are anodontia or oligodontia with malformations of teeth present in both deciduous and permanent dentition. The roots of the teeth are usually short and conical. In case of complete anodontia the alveolar process will fail to develop and the vertical dimension will reduce leading to protruberant lips.\(^3\)\(^8\) This paper reports two cases of ectodermal dysplasia in siblings without any family history.

Case Report 1

A 19 year old male came to department of oral medicine and radiology with the chief complaint of missing teeth. On extraoral examination it was observed that he had receding hair line, scanty eyebrows, dry and parched skin which gave a scaly appearance (Figure 1, Figure 2). He had depressed nasal bridge, prominent supra-orbital ridges and frontal bossing. His lips were protruberant. He gave history of having difficulty in tolerating high temperature especially during summers. On intraoral examination he had many missing teeth. The only teeth present were 17,16,13,23,27 (Figure 3). The canines were cone-shaped and molars had altered morphology. The mandibular ridge was completely edentulous and appeared flat (Figure 4). The overall vertical dimension of the face was reduced. Panoramic radiograph was taken to look for any impacted teeth. On radiographic examination no impacted teeth were found. The roots of the teeth present were found to be short and conical (Figure 5).

Case Report 2

A 16 yr old male patient, younger brother of previous patient reported to the department with chief complaint of missing teeth. On extraoral examination he had features similar to his brother with scantier scalp hair. On intraoral examination, he also had many missing teeth. The only teeth present were 16,11,21,26. All the teeth were conical in shape with altered morphology. The mandibular ridge was completely edentulous and appeared flat. The overall vertical dimension of the face was reduced. Panoramic radiograph was taken to look for any impacted teeth. On radiographic examination no impacted teeth were found. The roots of the teeth present were found to be short and conical (Figure 6-9). Based on history, clinical appearance of the patient, intraoral examination and radiographic investigations diagnosis of hereditary hypohidrotic ectodermal dysplasia was arrived at.

Discussion

Hypohidrotic ectodermal dysplasia was first described in 1848 by Thurnam, and later in the 19th century by Darwin. 9It was assigned to the X chromosome in 1921 by Thadani, who later reported that the carrier females could manifest signs of the condition.\(^8\) Christ in 1913, defined it as congenital ectodermal defect, Weech in 1929, impressed by the depression of sweat glands coined the term “anhidrotic ectodermal dysplasia.”\(^9\) Incidence HED is found in all racial groups and in all areas of the world. The incidence at birth was estimated by Stevenson and Kerr, based on the prevalence of HED in Oxfordshire: they suggested a rate of 1 per 100,000 births.\(^8\) Various classifications for this disorder exist. Freire-Maia and Pinheiro have proposed a classification based on the involved ectodermal derivative. This condition indicates hair dysplasia, dental dysplasia, nail dysplasia, and sweat gland dysplasia. Based on this there are more than 10 subgroups in Freire-Maia and Pinheiro classification.\(^2\) Clinically ectodermal dysplasia may be divided into two broad categories i.e., the hypohidrotic form which is X-linked, characterized by the classical triad of hypodontia, hypotrichosis and hypohidrosis, which is also termed as Christ Touraine Syndrome.\(^10\) The other category is that of hidrotic form described by Clouston which usually spares the sweat glands and can affect the teeth, hair and nails.\(^8\) The presen-
tation of facial deformity, dry skin, sparse hair of scalp, eye-
brows and eyelashes and hypodontia observed in these cases
is similar to previous reports.6,10-12 Dryness of the skin and ec-
zema are due to anomalies of the skin appendages which may
include partial or total absence of hair follicles, sweat glands
and sebaceous glands. The intolerance of heat and hyperther-
ia is also similar to previous reports which is attributed to
absence of sweat glands.2,6 The appearance of nails of both
fingers and toes was normal. This was in accordance with pre-
vious reports though contradictory to those of Akhyani.2,6
Xerostomia observed in present case was also similar to previous
reports which could be due to partial or complete absence
of salivary glands.6 Many previous reports have shown that
these patients have complete anodontia, i.e., missing decidu-
ous and permanent teeth, though both our patients had few
missing maxillary permanent teeth and completely missing
mandibular teeth.11,12 The teeth had altered morphology and
they appeared conical. Absence of teeth leads to reduced
growth of alveolar process. This in turn leads to reduced ver-
tical dimensions of occlusion and protuberant lips.6,10 Thus,
we report two cases of ectodermal dysplasia in siblings with
trichodysplasia, hypodontia and dyshidrosis. Thereby both the
cases reported come under 124 subgroup of the classification.
10Edentulousness can result in potentially stressful situation
that can influence the patient’s whole outlook in life. Prosthe-
sis either removable or fixed can be used to replace missing
teeth and enhance the patient’s confidence and personality.
The patient was referred to the department of Prosthodontics
for further management.

Conclusion
In conclusion Ectodermal dysplasias (EDs) are a heteroge-
eous group of disorders characterized by the triad of signs
comprising sparse hair, abnormal or missing teeth and inabil-
ity to sweat due to lack of sweat glands. When indicated the
dentists plan, appropriate care needs to be rendered to main-
tain oral functions as well as to address the aesthetic needs of
the patient.

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