Ectodermal dysplasia (ED) was first described by Thurman as a congenital, diffuse, non-progressive entity that occurs as a result of an ectodermal disturbance in the primitive stage of growth development [1]. Frequent phenotypic manifestation includes sparse hairs and deficiency of sweat glands. The common oral manifestations are anodontia or oligodontia, generalized spacing between teeth with malformation of teeth present in both deciduous and permanent dentition, and cleft lip and palate [2]. Other features include prominent supraorbital bridges, frontal bossing, depressed nasal bridges, and hyperthermia. The patient often has unexplained fever as a result of loss of sweat glands. Based on the number and functioning of sweat glands, ED is classified as hypohidrotic ectodermal dysplasia (HED) also known as Christ-Siemens-Touraine syndrome, which affects hair, nail, skin, and sweat glands and hidrotic ED, also known as Clouston syndrome which affects hair, nail, skin sparing the sweat glands [3].

**CASE REPORT**

A 12-year-old female came to the Department of Oral Medicine and Radiology with complaints of unerupted teeth and mastication difficulties. No similar condition was reported in her family members.

On examination, she had dry skin, sparse hairs and scanty eyebrows, broad supraorbital bridges, overfolding ears, saddle nose, protuberant lips with perioral pigmentation, and normal nails on extra-oral inspection (Fig. 1). Intraorally, several teeth were missing (13,14,15,22,24,25,27,35,34,33,32,31,41,42,44,45,47). A high arched palate was noted (Fig. 2).

On radiological examination, panoramic radiograph revealed unerupted 13,18,25,37 and 47 (Fig. 3). Jaw growth was not impaired. The alveolar process did not develop favorably and the vertical dimension was reduced. Trichodontosseous syndrome and partial anodontia were among the differential diagnosis. The case was provisionally diagnosed as ED based on clinical features, history of intolerance to high temperature, and radiographic examination.

The patient was motivated for prosthetic rehabilitation. Impressions were made and a removable partial denture has been delivered to the patient with the able guidance of prosthodontist and pedodontist (Fig. 4). This has improved her physiological and psychological status. She is on regular visits, and the patient was educated to avoid long-term activities and was explained about the genetic inheritance of this condition with appropriate counseling. Maybe in near future, she agrees to endosseous dental implants after the eruption of 13, 18,25 amd 47.

**DISCUSSION**

Freire-Maia defined the nosologic group of ED as a pathogenic developmental defect with any group that exhibits at least two of
the following features: Trichodysplasia (abnormal hair), abnormal dentition, onchodysplasia (abnormal nails), and dyshidrosis (abnormal or missing sweat glands) [4]. The developmental disturbance also causes partial or complete absence of permanent or primary dentition. The partial (oligodontia) or complete absence of teeth (anodontia) in both primary and permanent dentition is the most prominent feature. Apart from the number, the shape of the tooth is also altered, mainly conical in shape with that taurodontism is also seen in a few cases [5].

The clinical features include sparse, fine, lusterless hair with abnormal scalp texture, eyebrows and eyelashes, dry skin, nail defects, frontal bossing, depressed nasal bridge, and protuberant dry and cracked lips. Complete or partial absence of sweat glands can lead to dry skin and eczema, and there may be heat intolerance or hyperthermia in extreme weather conditions [5].

Mutations in genes that code for cell-cell communication, adhesion molecules, or transcription regulators may cause ED syndrome. The cause could be a mutation or deletion of specific genes, which can be inherited or happen in people who have no family history [6]. More than 300 cases studied genetically revealed that the X-linked mode of inheritance, which has its gene locus at Xq11–21.1, is carried by the female but exhibited only in males [7]. ED is genetically related and the genes that control are EDA (ectodysplasin), EDARADD (ectodysplasin-receptor associated death domain), and EDAR (ectodysplasin receptor). ED can be autosomal dominant, autosomal recessive, or X-linked recessive [8]. Earlier research studies have found that mutations in the gap junction beta 6 (GJB6) gene, which codes for the protein connexin30 (CX30) are the most common cause of HED [9]. HED is characterized by syndromic tooth agenesis caused mostly by mutations in the ectodysplasin A (EDA), ectodysplasin A receptor (EDAR), EDARADD, and WNT10A genes. The triad symptoms of HED include hypohidrosis, hypotrichosis, and hypodontia [10]. Normal intelligence is observed [11]. Genetic alterations of ED-associated genes that only affect one derivative of ectoderm (e.g., hair, teeth, nails, and sweat glands) should be grouped as non-syndromic traits of the causative gene (e.g., non-syndromic hypodontia or missing teeth associated with pathogenic EDA variants) [12].

The patient in our study was not subjected to genetic analysis due to money constraints.

The most striking features in this case report are that the patient belong to HED and has a history of intolerance to hot environmental exposure with no genetic inheritance. She had dry skin, sparse hair and eyelashes, normal fingernails, and toenails, partial anodontia with altered tooth morphology. Missing teeth were more in the mandible than in the maxilla with maxillary hypoplasia. Skin biopsy and sweat pour test can be done to rule out hypohidrosis [13]. She was managed with a removable partial denture to create a positive self-image look which has improved her psychological status. She has planned for endosseous dental implants after the eruption of 13,18,25 and 47 teeth.
CONCLUSION

The physical and mental health of a person is affected by the clinical signs of ED. Through a multidisciplinary approach, the patient’s speech, esthetics, and ability to eat a diverse diet have all improved and have the patient’s acceptance among peers and family.

REFERENCES


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