Case Report

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An Insight of Ectodermal Dysplasia in a Case Report

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ABSTRACT

Background: Ectodermal dysplasia is an extremely rare hereditary disease characterised by dysplasia of ectodermal origin tissues primarily nail, teeth and skin or sweat glands. Common manifestations include defective hair follicles and eyebrows, nasal bridge depression, protuberant lips, anodontia or hypodontia and conical teeth. Dry skin, hyperthermia and high fever may also occur due to deficiency of sweat glands. It generally affects the male more than female and occurs mostly during first or second decade of life. There is no specific treatment for ectodemal dysplasia, only disease management are done in such cases. Here we present a case of 15 year old male patient who exhibited many manifestations of ectodermal dysplasia.

Keywords: Ectodermal dysplasia, anodontia, Christ-Siemens-Touraine syndrome, Clouston syndrome.

INTRODUCTION

Ectodermal dysplasias (EDs) are a heterogenous group of disorders initially described by Thurnam who was the first to report and publish a case of ED in 1848[1]. They are characterized by developmental dystrophies of ectodermal structures namely hypohydrosis, hypotrichosis, onychodysplasia and hypodontia or anodontia[2]. Disturbance in the development of ectoderm in the embryo leads to this condition[3]. Till date more than 150 distinctive syndromes have been described with all possible modes of inheritance, commonest being Hypohydrotic (Anhydrotic) ED and Hydrotic ED[4].

Hypohydrotic form otherwise known as Christ-Siemens-Touraine syndrome exhibits hypohydrosis (reduced sweating), hypotrichosis (sparse hair) and hypodontia (reduced number of teeth). Males are affected severly as it is a X-linked recessive inheritance[5]. Teeth, hair and nails are affected in the hydrotic form of ED where as the sweat glands are usually spared. It shows an autosomal dominant trait but autosomal recessive inheritance is also reported[5].

Various Dental anomalies of ectodermal dysplasia are complete anodontia or hypodontia, abnormal shape of teeth like conical or pegged teeth and delayed eruption of permanent teeth[4]. This may lead to underdevelopment of the alveolar ridge[6]. Various other manifestations like saddle nose, prominent lips, linear wrinkles and hyperpigmentation around the eyes, facial dysmorphic features are also associated with it[7]. The patients with such cases are treated with team effort of specialists from dental surgeon to plastic surgeons to restore the functional and aesthetic corrections of the face. Here we report a case of hypohydrotic ectodermal dysplasia which was encountered in our institution.

CASE REPORT

A 15 year old male patient reported to the department of Oral Medicine and Radiology at Kalinga Institute of dental Sciences with a chief

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complaint of missing teeth from past 6 years. Patient gave a history of normal eruption and shedding of deciduous teeth. After shedding of deciduous teeth they were not replaced by their permanent successors. The patient had difficulty in mastication and speech. The patient also complained of reduced sweating and repeated episodes of unexplained fever. No relevant medical, family or personal history was elucidated.

On extra-oral examination, patient presented bilaterally symmetrical long narrow face, thin sparse and coarse scalp and body hair, frontal bossing, sparse eyebrow hair and increased intercanthal distance. Areas of hypopigmentations were seen in the zygomatic region bilaterally. Patient presented with a thick protruding everted upper lip and prominent chin (Fig.1). Wrinkled skin periorbitally giving an old man look was seen (Fig.2).

Fig 1: Everted upper lips.

Fig 2: Periorbitally giving an old man look

Intraoral examination revealed partial anodontia of the permanent dentition with presence of only 3 teeth (11, 21, 14) in the oral cavity of which the anterior teeth were conical in shape. Complete absence of mandibular teeth was seen (Fig.3).

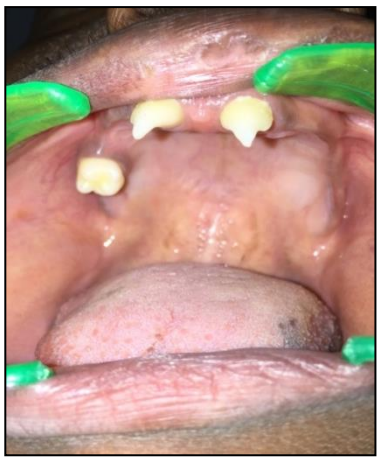

Fig 3: Complete absence of mandibular teeth.

All routine investigations were normal. CBCT image confirmed the partial anodontia status and showed only 3 permanent teeth in the maxillary arch with absence of mandibular teeth (Fig. 4). The lateral skull view showed class III skeletal tendency with reduced lower facial height (Fig. 5). Based on the clinical and radiographical findings, a final diagnosis of Ectodermal Dysplasia was made. A removable partial denture for the maxillary arch and complete denture for the mandibular arch was provided as the treatment options with composite resin modifications for the conical shaped maxillary anteriors.

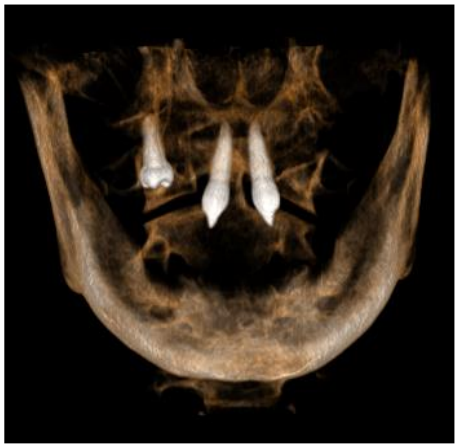

Fig 4: Complete absence of mandibular teeth.

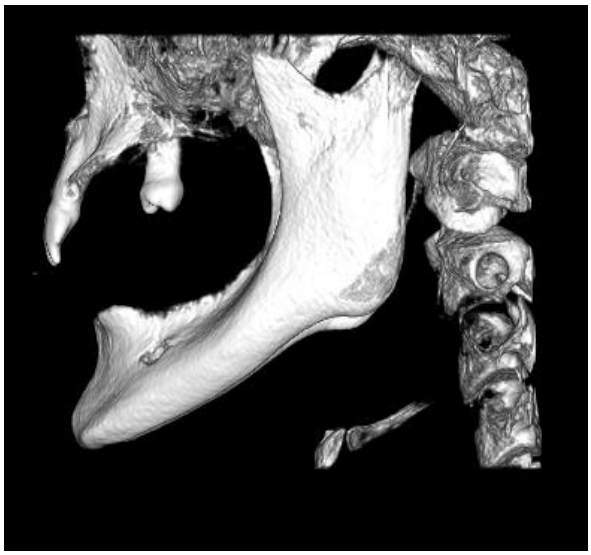

Fig 5: Class III skeletal tendency with reduced lower facial height

DISCUSSION

Ectodermal dysplasia is a genetic disorder, X linked mode of inheritance occurring in the gene locus Xq12-q13.1. The gene is usually carried by the females but mostly manifests in males [8]. Ectodermal dysplasia is of different types and the signs and symptoms also markedly different depending upon the structures or organs involved i.e hair, nails, teeth and sweat glands. The signs and symptoms may not be distinct in newborns or childhood [9]. The most common finding in such patients is the reduced number of teeth and their deformed shape and size which helps in its diagnosis. The patient in the present case also had few teeth with conical and small maxillary anteriors.

Patients with ED show different ectodermal structures defects of which different combinations are usually encountered such as hair and nails defect in some individuals where as teeth and sweat glands [10]. In the present case we found hair and teeth defects as the combination where thin sparse hair on scalp and eyebrows were encountered on examination. Protruberent and everted lips, pointed chin with prominent supraorbital ridge which are some of the important extraoral characteristics observed in ED were also evident in the present case. Skin biopsy and genetic testing are some of the diagnostic tests for Ectodermal dysplasias.

Some of the commonly occurring syndromes are Clouston syndrome, Hay-Wells syndrome, Ectrodactyly-ectodermal defects (EEC) syndrome, Rapp-Hodgkin Ectodermal Dysplasia and Hypohidrotic Ectodermal Dysplasia which shows symptoms like palmoplantar hyperkeratosis, ectrodactyly and syndactyly of the hands, cleft lip etc [9].

The molecular pathogenesis of ED is not properly understood till yet. Ectodysplasin-A, the protein which is necessary for the normal development of ectodermal appandages are produced by the gene involved in ED. So any disturbance or defects in this ectodysplasin-A may inhibit the normal development of the ectoderm [11,12].

Patients with ED need utmost care and revisions which involves physical, psychological as well as oral rehabilitations. As there is no pharmacological management of these patients, the treatment is mostly based on the organ involved. The patients are usually advised to use artificial tears and skin moisturizers, avoid of excessive exposure to warm or dry climates with heavy physical exercises [13].

Dental treatment is necessary for such patients to restore mastication and speech for which prosthesis and aesthetic dental interventions helps to improve the overall oral health. Dental implants or implant supported dentures depending upon the underlying bone density and height are recommended as the treatment choice. Regular follow-up with psychological assistance and care helps and facilitate social acceptance in patients suffering from ED.

CONCLUSION

In developing country like India, patients with Ectodermal dysplasia are generally ignored socially for which they are being diagnosed with many associated systemic and psychological disease. Genetic disease has no absolute cure or treatment but symptomatic management of such disorders can be done for better aesthetic which can help these patients to live similar life like others in the society.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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