ECTODERMAL DYSPLASIA (HYPOHIDROTIC SYNDROME) WITH COMPLETELY EDENTULOUS MANDIBLE

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Abstract

Ectodermal dysplasia is a hereditary disease characterized by congenital dysplasia of one or more ectodermal structures and other accessory appendages. It usually affects the males, and females are the carriers. The clinical features include sparse hair and inability to sweat due to lack of sweat glands. Dental manifestations include hypodontia, complete anodontia or malformed teeth. Oral rehabilitation is the major challenge in such patients. A case report 38 years old male patient with characteristic clinical features of hypohidrotic ectodermal dysplasia is reported. Intraoral examination revealed partial anodontia of teeth. He was the only member of his family who suffered from hypohidrotic ectodermal dysplasia.

Key words: Anodontia, Christ-Siemens-Touraine syndrome, Hidrotic, Hypohidrotic, Trichondysplasia.

Introduction

Ectodermal dysplasia (ED) is defined by National foundation for ectodermal dysplasia as a genetic disorder in which there are congenital birth defects of two or more ectodermal structures (Hickey, 2001). These tissues primarily are the skin, hair, nails, exocrine glands, and teeth. Defects in tissues derived from other embryologic layers are not uncommon. The disorders are congenital, diffuse, and nonprogressive. Thurman published the first report of a patient with ED in 1848,[1] but the term was not coined until 1929 by Weech.[2] Freire-Maia and Pinheiro described numerous varieties of ectodermal dysplasia involving all possible Mendelian modes of inheritance.[3] The ectodermal dysplasias are congenital, diffuse, and nonprogressive. The most common ectodermal dysplasias are X-linked recessive hypohidrotic ectodermal dysplasia (Christ – Siemens – Touraine syndrome), and hidrotic ectodermal dysplasia (Clouston syndrome).[4] Ectodermal dysplasia patients require both functional and aesthetic corrections of the face. The absence of alveolar bone and teeth is a difficult reconstructive challenge for the surgeon.

Case Report

A 38 year old male patient reported to the Department of Oral Medicine and Radiology with the chief complaint of lack of esthetics, difficulty in mastication. The patient also complained of heat intolerance, but was bothered mainly due to the missing teeth. He was the only member of his family no similar history in the family. Past medical and treatment history was insignificant. No positive family history was found. On general physical examination, the patient had frontal bossing and sunken cheeks with thick everted lips. Periorbital hyperpigmentation was present. Hair was found to be very fine and brittle hair on the scalp. (Figure 1)

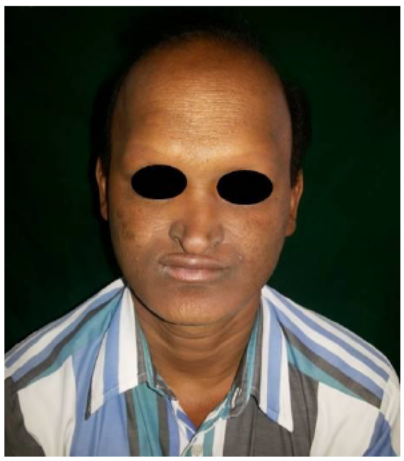

Figure 1: Profile view show typical facial features of Ectodermal Dysplasia (ED).

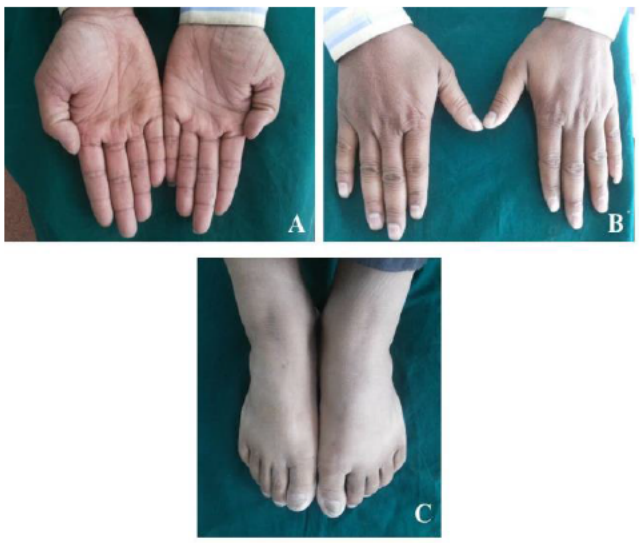

Figure 2: A) Dry surface of palm; B) Nail deformities with longitudinal ridges. C) Dry skin and nail deformities of leg.

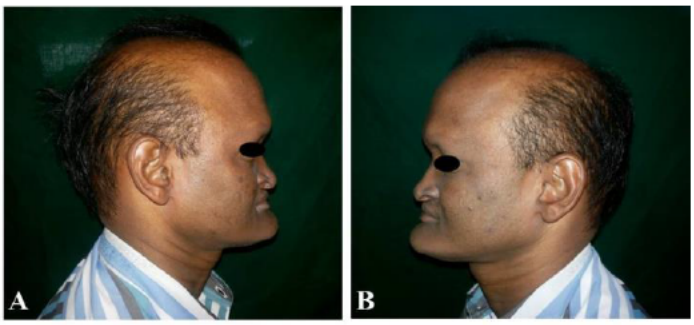

Figure 3 A: Lateral view shows sunken and deficient maxillary complex and prognathic mandible generally seen in edentulous geriatric patients

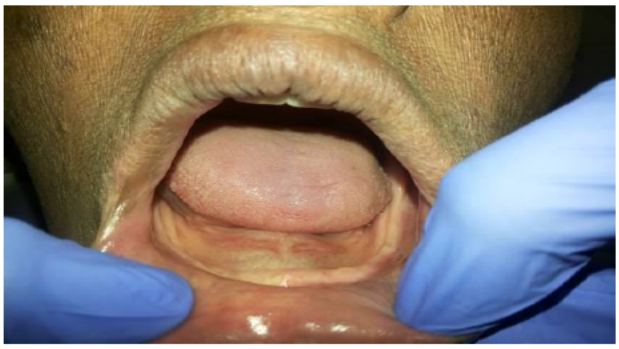

Figure 3 B: Intraoral photograph showing complete edentulous mandible

Underdeveloped maxillary alveolar ridges is present. Partial anodontia of maxilla and edentulous mandibular teeth and underdeveloped alveolar ridge

All routine investigations were normal. OPG (Orthopantogram) (Figure 4) and lateral cephalogram (Figure 5) was performed, which confirmed the absence of alveolus and the other teeth are congenitally missing. The combined dental and clinical findings pointed towards a diagnosis of hypohidrotic ectodermal dysplasia (Christ-Siemens-Tourine syndrome). The treatment option preferred was dental implants , as it would have given him long term results of better mastication, preservation of alveolar bone and better functional esthetics than removable prostheses (Dentures). Due to financial constraint patient opted for dentures.

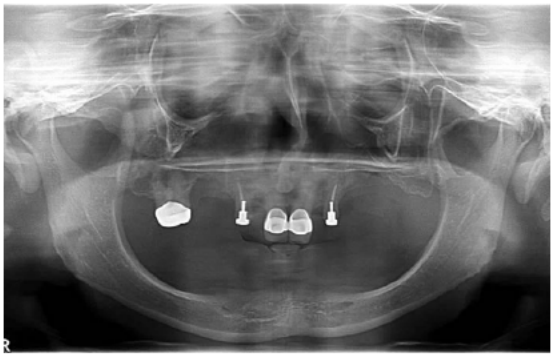

Figure 4: Orthopantogram of patient.

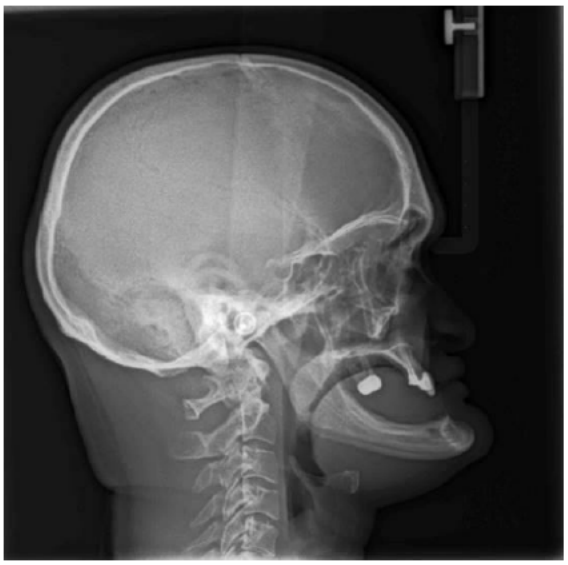

Figure 5: Lateral Cephalogram of patient

Discussion

The ectodermal dysplasias comprise a large, heterogenous group of inherited disorders that are defined by primary defects in the development of two or more tissues derived from the embryonic ectoderm. The condition is thought to occur in approximately one in every 100,000 births.[5] Genetic studies of more than 300 cases have revealed X linked mode of inheritance with its gene locus being Xq11-21.1, the gene is carried by the female but manifested in the male.[6] However there are reports of multiple siblings being affected and of females suffering with this condition. The first classification system for ectodermal dysplasias was proposed by Freire-Maia and Pinheiro in 1982,[7] with additional updates in 1994 and 2001.[8]

The patients were stratified into subgroups based on presence or absence of the following: [9]

1. Trichondysplasia (abnormal hair)
2. Abnormal dentition
3. Onchondysplasia (abnormal nails)
4. Dyshidrosis (abnormal or missing sweat glands)

Overall, the ectodermal dysplasias were classified into either group A disorders, which were manifested by defects in at least two of the four classic ectodermal structures as defined above, with or without other defects, and group B disorders, which were manifested by a defect in one classic ectodermal structure (1-4 from above) in combination with (5), a defect in one other ectodermal structure (i.e., ears, lips, dermatoglyphics). Eleven group A subgroups were defined, each with a distinct combination of two or more ectodermal defects (e.g., 2-4, 1-3, 1-4 from above). The group B disorders were indicated as 1-5, 2-5, 3-5 or 4-5 (from above).

Clinically, hereditary ectodermal dysplasias may be divided into two broad categories:

1. X-linked hypohidrotic form (Christ-Siemens-Tourine syndrome), characterised by the classical triad of hypodontia, hypohidrosis and hypotrichosis and by characteristic dysmorphic facial features. Our patient was suffering from this syndrome, which presents with the following features.[10,11]
    a) The typical faecies, which is often not recognised until infancy, is characterized by frontal bossing, sunken cheeks, saddle nose, thick, everted lips, wrinkled, hyperpigmented periorbital skin and large, low-set ears. (Figure 3A)
    b) Dental manifestations include conical or pegged teeth, hypodontia or complete anodontia and delayed eruption of permanent teeth. (Figure 3B)
    c) Most patients have fine, sparse, lustreless, fair hair; therefore, little pigmentation in the hair shaft is observed microscopically and the medulla is often discontinuous. When medullation is present, a "bar code" appearance is often seen.
    d) Onychodystrophy may occur, but is not common.
    e) Extensive scaling of the skin and unexplained pyrexia secondary to anhidrosis may occur in the neonatal period. The development of a chronic eczematous dermatitis is common.
    f) Other common signs are short stature, eye abnormalities, decreased tearing and photophobia.
2. Hidrotic form (Clouston's syndrome) that usually spares the sweat glands but affects the teeth, hair and nails. Most of the other clinical features are similar to that seen in the hypohydrotic form. It has an autosomal-dominant inheritance and is common in persons of French-Canadian ancestry[12,13] Nail dystrophy associated with hair defects on palmoplantar dyskeratosis. Nails are thickened & discoloured, scalp hair sparse, fine and brittle.

Differential Diagnoses

- Alopecia Areata
- Aplasia Cutis Congenita
- Focal Dermal Hypoplasia Syndrome

Investigations

In general, laboratory studies are not useful in the diagnosis or management of the ectodermal dysplasias. Patients with ectodermal dysplasia associated with immunodeficiency may have hypogammaglobulinemia with impaired lymphocyte proliferation and cell-mediated immunity. An appropriate evaluation, including determination of quantitative immunoglobulin levels and T-cellsubset populations, should be performed. Perform orthopantography at an early age if hypodontia or dental abnormalities are present. X-ray films of hands, feet, or both may demonstrate specific skeletal deformities. Sweat pore counts, pilocarpine iontophoresis, and skin biopsy may document hypohidrosis and a reduction in the number of eccrine glands.

Treatment

Dental treatment is often necessary in patients with some forms of ED and some children may need dentures as early as 2 years of age. It is important to seek dental advice early as maintenance of the alveolar ridge is important for later dental intervention. Prosthetic teeth are implanted in adults for mastication and speech. Importantly, aesthetic dental interventions in patients with ED and malformed teeth and malocclusion helps with the development of a positive self-image and overall oral health.

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