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ORIGINAL ARTICLE

Christ-Siemens-Touraine Syndrome - A Case Report with Review of literature

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Abstract:

Ectodermal dysplasia (ED) is a hereditary disorder characterized by developmental defects of structures derived from the ectoderm. The most prevalent form of ED is hypohydrotic ectodermal dysplasia also known as Christ-Siemens-Touraine syndrome. This case report illustrates the manifestations and diagnosis of a twenty-two-year-old male patient with hypohydrotic ectodermal dysplasia with a brief review of literature.

Keywords: Syndromic oligodontia, ectodermal dysplasia, hypohidrotic ectodermal dysplasia

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INTRODUCTION

Oligodontia is defined as the congenital absence of six or more teeth in the dentition, excluding the third molars. Oligodontia can be classified as syndromic or nonsyndromic. Nonsyndromic oligodontia is a developmental dental anomaly without involvement of other organs. Syndromic and nonsyndromic forms of oligodontia can be differentiated by conducting a thorough physical examination of the hair, nails, sweat glands, eyes, and by genetic tests for any congenital disorders. Syndromes associated with oligodontia are enumerated in Table 1.[1,2,3] Ectodermal dysplasia (ED) is a hereditary disorder caused by disturbances in the ectoderm of the developing embryo. Here we report a case of oligodontia in a patient with the hypohidrotic type of ED along with a review of literature.

Table 1. Syndromes associated with Oligodontia

Ectodermal Dysplasia
Incontinentia Pigmenti
Down Syndrome
Rieger Syndrome
Wolf-Hirschhorn Syndrome
Van Der Woude Syndrome
Oral Facial Digital Syndrome
Witkop Tooth And Nail Syndrome
Klinefelter Syndrome
Ellis Van Creveld Syndrome

CASE REPORT

A twenty-two-year-old male patient came to us with the complaint of missing upper and lower teeth. The patient gave history of difficulty in speech and eating. He also gave history of decreased sweating and sparse hair growth since childhood. He was unable to tolerate hot weather and visited a doctor for the same. The patient was explained he had symptoms of a genetic defect. No investigation or treatment was done. There was no history of repeated fever, dryness of mouth or lack of tears. Family history revealed consanguineous marriage of parents. He had an older brother with the same condition.

The patient was moderately built and nourished. Extra – oral examination revealed frontal bossing, depressed nasal bridge, obliquely placed ears, protuberant lips, sparse fine hair on scalp, normal beard growth, and decreased vertical dimension. (Figure 1) His eyes appeared sunken with faint eyebrows and the absence of eyelashes. (Figure 2) His skin appeared smooth and dry with no changes seen on the hands or nails. (Figure 3)

Intra oral examination revealed two conical central incisors in the maxillary arch. The mandibular arch was edentulous with resorbed knife-edge residual alveolar ridges. (Figure 4)

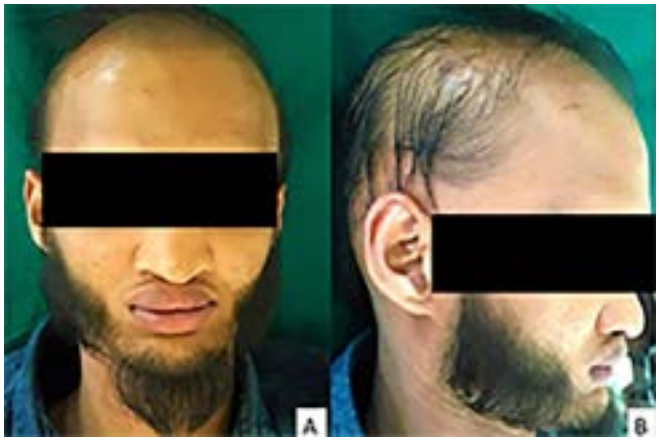

Figure 1. Facial profile (A) Frontal and (B) Lateral showing extra -oral features of ED

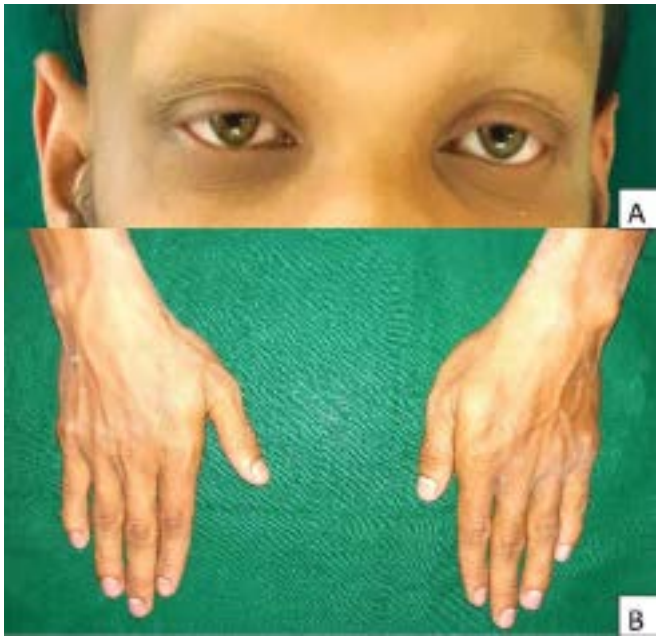

Figure 2. Clinical photograph showing (A) sunken eyes, absence of eyelashes and eyebrows

A digital panoramic radiograph (OPG) was advised to check for the presence of permanent tooth buds. The OPG revealed bilaterally normal condylar and coronoid processes with normal appearance of jaws and trabeculae. Two conical shaped teeth were seen in the maxillary anterior region. The mandibular arch was edentulous. There was no evidence of permanent tooth buds.

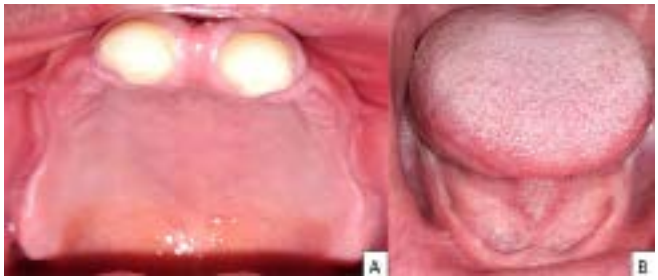

Figure 3. Intraoral photograph of (A) Maxillary arch showing two conical incisors (B) Mandibular arch showing knife edge residual ridge and the absence of teeth

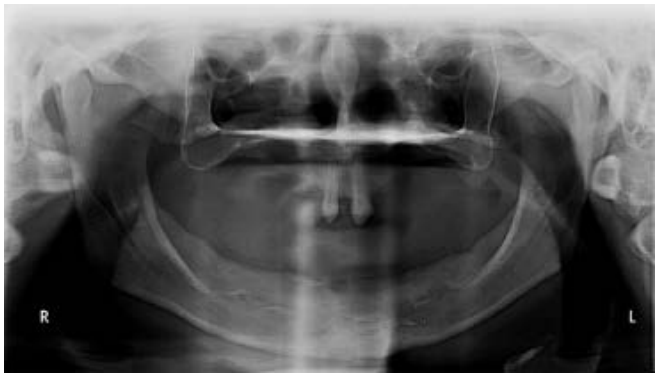

Figure 4. OPG showing the presence of two conical incisors and the absence of any permanent tooth buds

The case was diagnosed as Hypohidrotic Ectodermal dysplasia. The patient was referred to the Department of Prosthodontics for rehabilitation.

DISCUSSION

ED was first reported in 1792 by Danz. In 1838, Wedderburn documented ED in a letter to Charles Darwin describing ten cases of Hindu male family members. In 1848 Thurman reported 2 male first cousins and their maternal grandmother with a hereditary syndrome associated with sparse hair, missing teeth and dry skin. The term ED itself was coined in 1929 by Weech.[4,5]

Occurrence of ED is approximately 1 in 1,00,000 live births with a mortality rate of 28% in males up to 3 years of age. It is more prevalent in males than females, with a ratio of 1.7:1. There are more than 190 types of ED. The two major forms are hypohidrotic ED and hidrotic ED.[6] Felsher in 1944 changed the adjective anhydrotic to hypohydrotic because the persons termed as anhydrotic were not truly devoid of sweat glands.[7]

The hypohidrotic form is also known as Christ-Siemens-Touraine syndrome. It represents 80% cases of ED. It is characterised by a triad of hypodontia, hypohidrosis and hypotrichosis as seen in the reported case. The hidrotic form is also known as Clouston's Syndrome. It shows features such as nail dystrophy, alopecia, and palmoplantar hyperkeratosis.8

ED is an X linked recessive condition. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. Mutations in the EDA, EDAR, and EDARADD genes cause hypohidrotic ectodermal dysplasia. These genes code proteins during embryonic development which form part of a signaling pathway that is critical for the interaction between two cell layers, the ectoderm and the mesoderm. There is an increased predisposition in consanguineous marriages.[9,10] Our patient is the offspring of a consanguineous marriage. He has an elder brother with the same condition. This supports the genetic etiology of the disease.

Ectodermal dysplasia is diagnosed by physical examination. Determination of quantitative immunoglobulin levels and T-cell subset populations can be performed to evaluate hypogammaglobulenemia associated with ectodermal dysplasia. Radiographs can be taken to rule out dental abnormalities. Genetic testing for mutations may be done. Other tests include biopsy of the hypothenar eminence, sweat pore count, Blaschko lines and pilocarpine iontophoresis. Prenatal testing by fetal skin biopsy also facilitates intrauterine diagnosis.[11,12,13]

There is no specific treatment for ED; only disease management is possible. Dental prosthetics, oral implants and supplementary bone augmentation techniques have been carried out in patients with oligodontia. Allogeneic stem cell transplantation has been performed in a small number of patients with autosomal dominant ED with immunodeficiency; poor engraftment and post-transplant complications were common.[14,15,16,17]

CONCLUSION

Patients with ED not only suffer from poor physiological development but also poor psychological development as a result of unacceptable esthetics. Timely diagnosis, counselling and rehabilitation is crucial to enable them to lead a normal life. Since anodontia/hypodontia greatly effects esthetics, such patients are likely to visit a dentist early in life. In such a situation, an oral physician may be the first to diagnose ED.

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