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CASE REPORT

CHRIST-SIEMENS-TOURAINE SYNDROME: A CASE REPORT

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A B S T R A C T

Christ–Siemens–Touraine syndrome is an anhidrotic or hypohidrotic form of ectodermal dysplasia (ED) characterized by triad of hypodontia, hypotrichosis, and hypohidrosis. Here we report a typical case of ED associated with impacted mandibular incisors and considering an interdisciplinary approach for its management.

INTRODUCTION

Ectodermal dysplasia (ED) syndrome was first described by Thurnam (Thurnam, 1848) and the term ED was coined by Weeck later in 1929.

National foundation for ED defines it as a group of genetic disorders which manifests as congenital birth defects affecting two or more ectodermal appendages. These disorders are believed to be non-progressive and diffuse, with a mode of Mendelian inheritance. EDs are broadly categorised in two groups: (1) The hidrotic form known as Clouston syndrome and (2) anhidrotic or hypohidrotic form known as Christ–Siemens-Touraine syndrome. These disorders are considered to be relatively rare with an estimated incidence of 1 case per 100,000 live births.

Christ-Siemens-Touraine syndrome can be diagnosed clinically, manifesting as sparse body hair or eyelashes, absent or reduced sweating, absent to abnormally shaped tooth along with specific facial features like prominent supraorbital ridge, midface deficiency, wrinkled and hyperpigmented skin etc (Mokhtari and Lotfi, 2012).

CASE REPORT

A 12 year old male reported to the Out Patient Department with the chief complaint of spacing between the teeth and conical shaped teeth in the upper and lower front region of jaw. Patient's mother informed that the delivery was full term and normal with no complications or any history of trauma. Eruption of teeth was delayed and the erupted teeth were malformed and often caused trauma to the lower lip while eating.

Extraoral examination of patient revealed a mesoprosopic facial form and a convex facial profile with prominent supraorbital ridges, scanty eyebrows, saddle nose and competent lips. Further clinical examination revealed dry skin of face and hands with fine wrinkles around the eyes, sparse body hair and parrot beaked shaped finger nails.(Figure 1,2,3,4,5) Whereas intraoral examination revealed partial anodontia, only four conical shaped teeth in maxillary anterior region with spacing between them and first and second molars were present in the upper arch, in lower arch canines, first and second molars were present. (Figure 6,7,8)

Radiographic examination revealed two impacted incisors in the mandible anterior region as seen on the OPG (Figure 9), the impacted incisors were seen fully covered with a thick layer of bone. On the basis of these clinical findings a diagnosis of Christ-Siemens-Touraine Syndrome was made.

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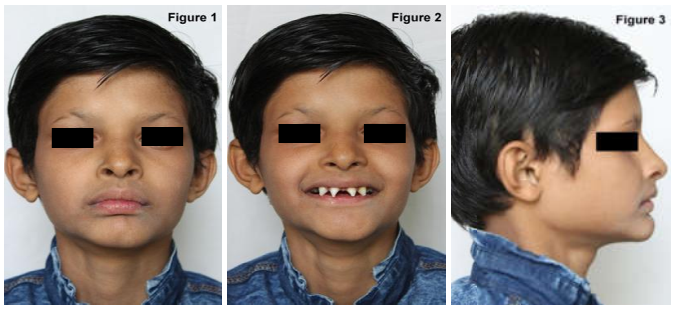

Figure 1,2,3 Extraoral Examination showing convex profile, scanty eyebrows, prominent supraorbital ridges and thick competent lips.

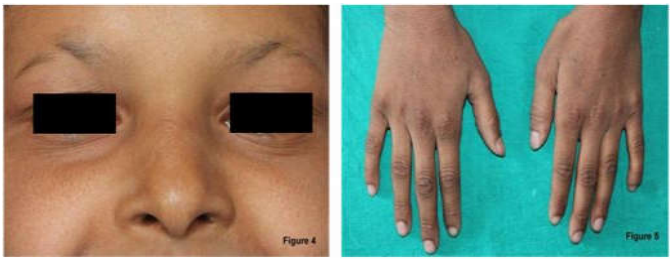

Figure 4,5 Dry skin of face with fine wrinkles around the eyes and dry skin of hand with parrot beaked shaped nails.

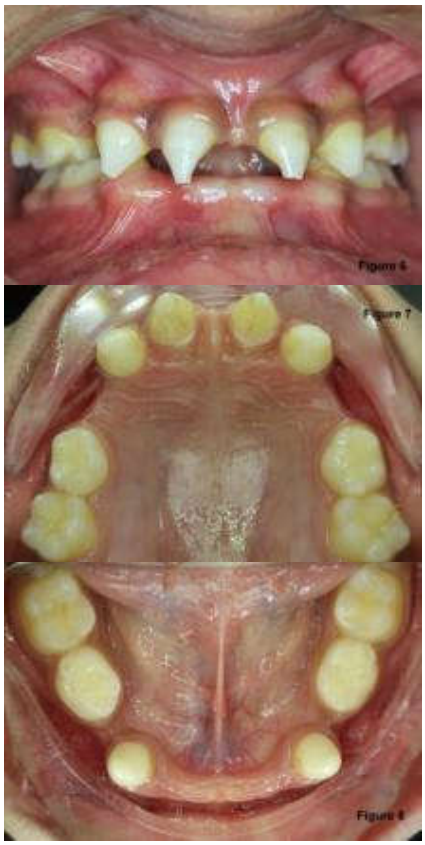

Figure 6,7,8 Intraoral Examination shows conical shaped teeth and missing teeth.

Causes

Christ-Siemens-Touraine syndrome can be inherited as an X-linked recessive, autosomal dominant and autosomal recessive disorder. The X-linked recessive HED (Hypohydrotic Ectodermal Dysplasia) is caused by mutation in EDA, which encodes for the ectodysplastin protein (Kere, 1996; Clarke *et al*, 1987). Whereas the autosomal dominant and autosomal recessive HED are caused by mutation in the DL gene, which encodes for the EDA receptor (Sandhu *et al*, 2007).

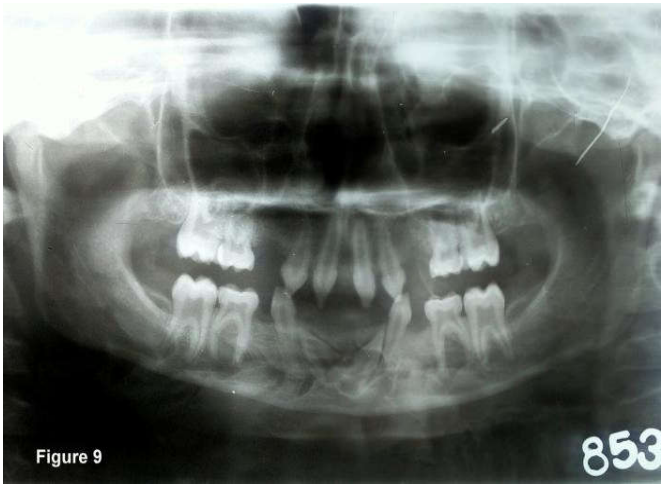

Figure 9 OPG showing impacted mandibular incisors

The other form of ED (Clouston syndrome or Hidrotic Ectodermal Dysplasia) which has an autosomal dominant mode of inheritance is caused by mutation in GJB gene (Mehta *et al*, 2007).

DISCUSSION

Different subgroups of EDs are created according to the presence or absence of the four primary ED defects: ED1: Trichodysplasia, ED2: Dental dysplasia, ED3: Onychodysplasia, ED4: Dyshidrosis (Goodman, 1985). Based on these primary ED defects, 192 different types of EDs are grouped into one of these subgroups.

Christ–Siemens–Touraine syndrome, subgroup 1-2-3-4, is characterized by the classical triad of hypodontia, hypotrichosis, and hypohidrosis (Pinheiro and Freire-Maia, 1979). Diagnosis is based primarily on the clinical grounds and whereas other modalities include Sweat pore counts, pilocarpine iontophoresis; radiographic evaluation of various anomalies; prenatal diagnosis using the genetic mutation analysis and biopsy of the mucus membranes and skin (Sybert, 1988).

The most common clinical findings include partial or complete absence of sweat glands resulting in decreased or no sweating, sparse scalp hair, scant or absent eyebrows, the eyelashes may be normal, sparse, or completely absent. Teeth abnormalities range from oligodontia to complete anodontia, abnormally shaped teeth with history of delayed eruption (Clarke *et al*, 1987). The nails are usually normal in most cases whereas some cases may present deformed nails. Craniofacial features include flat forehead with frontal bossing, concave facial profile, prominent supraorbital ridge, saddle nose, everted lips or double lip appearance, and periorbital hyperpigmentation and fine wrinkling around the eyes (Mehta *et al,* 2007). In our case most of these characteristic findings were present. Besides these, impacted lower incisors were seen on panaromic X-ray.

Treatment Plan

Managment of ED depends on the structure involved. In our case report, the patient was found to be having dry skin, sparse eyebrows and eyelashes, therefore he was advised to visit a dermatologist for the needful. For the management of dental defects, an interdisciplinary approach was taken into consideration, consisting of initial reshaping and crown build up of the conical incisors and canines in upper arch followed

by orthodontic alignment of the lower impacted incisors, reshaping and crown build-up of lower incisor and canines and closure of remaining space in between the incisors and canines after crown build-up in both the arches, followed by an upper and lower removable partial denture prosthesis to restore the functional and masticatory efficiency. After construction of dentures patient was educated for adjustments and reconstruction of dentures at different stages of growth and advised that dental implants may eventually be required. No dietary restrictions were indicated. The prognosis of the ectodermal dysplasia is quite good and the life span of the patient is usually normal except for case of ectodermal dysplasia with immunodeficiency or with other systemic manifestations.

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