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CHRIST SIEMENS TOURAINE SYNDROME-A CASE REPORT

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ABSTRACT

Christ-Siemens-Touraine (CST) syndrome, also referred to as hypohidrotic ectodermal dysplasia, belongs to a group of genodermatosis known as ectodermal dysplasia(ED). From the clinical point of view two main forms have been distinguished -**Hypohidrotic form/Christ-Siemens-Touraine Syndrome and Hidrotic type/Clouston syndrome**. The disorder hypohidrotic ectodermal dysplasia affects 1-7 per 100,000 live births. This disorder is usually inherited as either autosomal dominant / recessive or X-linked recessive trait. It is commonly X-linked recessive with full expression in males. Female carriers have a minimal expression. The hypohidrotic form is characterized by a classical triad of signs comprising hypotrichosis, hypodontia/anodontia, and anhidrosis/hypohidrosis. The X-linked recessive Ectodermal dysplasia is the most common disorder and constitutes 80% of EDs. This case report describes the multi-specialty oral rehabilitation of an 08 year old female patient with ectodermal dysplasia with investigations like radiographic and sweat pore count.

KEY WORDS: *Ectodermal dysplasia; Christ Siemen Touraine syndrome; X-linked Hypohidrotic Ectodermal Dysplasia*

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).[1,2] Female carriers outnumber affected men but females show little or no signs of the condition. Usually the ectodermal dysplasia is divided into two types based on the number and function of sweat glands as mentioned below:

1. Hypohidrotic form/Christ -Siemens-Touraine Syndrome
2. Hidrotic type /Clouston syndrome (1)

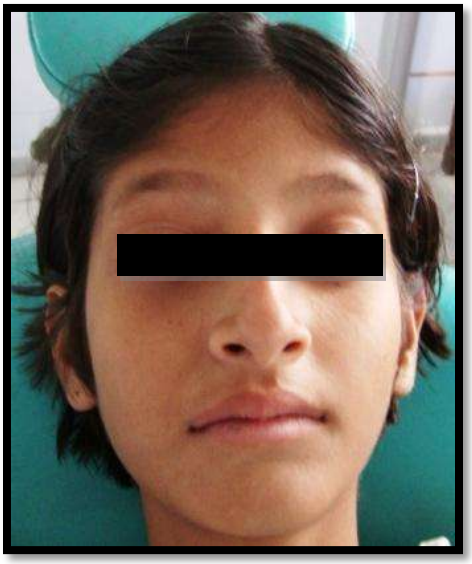

Fig 1 Pre-operative patient's photograph

Christ-Siemens- Tourine Syndrome - In this form sweat glands are absent or significantly decreased. This disorder is usually inherited as either autosomal dominant / recessive or X-linked recessive trait and the gene locus is X q13-q21. It is commonly X-linked recessive with full expression in males. Female carriers have a minimal expression. Usually 60-70 % of cases show manifestations restricted to minimal hypodontia, aplastic or hypoplastic mammary glands, impaired lacrimal gland function, glaucoma and increased susceptibility to allergic disorders such as asthma or eczema. Typical general mental development, frontal bossing with characteristics reduction in amount of hair (hypotrichosis), absence of sweat glands (anhidrosis) resulting in temperature elevation, absence of sebaceous glands (asteatosis) resulting in dry skin, depressed nasal bridge, protuberant lips, prominent supra orbital ridges , sunken cheeks, wrinkled hyperpigmented skin around the eyes and large low set ears.[1,2,3,4,5]

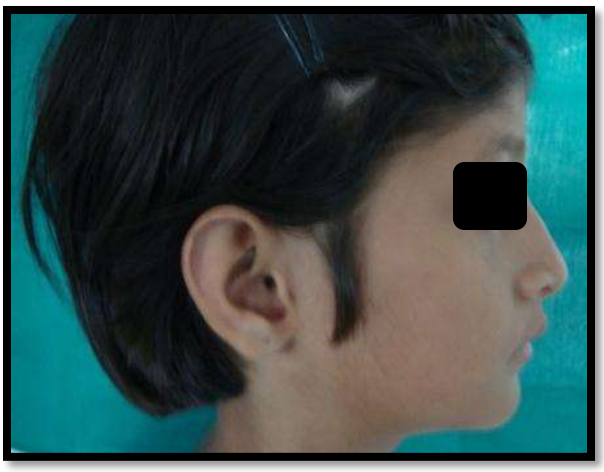

Fig 2: Pre-operative patient's photograph

CASE REPORT

An eight year old girl reported to the Department of Paedodontics and Preventive dentistry, Chandra Dental College & Hospital, Safedabad, Barabanki, U.P. with a chief complaint of missing front upper and lower teeth and difficulty in chewing food. General physical examination revealed sparse hair on the eyebrows (hypotrichosis), dry skin and lips, prominent supraorbital ridges and large low set ears [Figures 1&2].Child was moderately built and also gave history of heat intolerance (hypohidrosis). Oral examination revealed hypodontia in the maxilla and mandible with underdeveloped alveolar ridges [Figure 3]. Teeth present were **# 11 54 55 16 21 65**

26 73 74 36 81 82 43 85 46 and missing were **12 13 22 23 64 31 32 84.** Apart from this, midline diastema and fractured left maxillary central incisor was seen. Family history was non-contributory.

PROVISIONAL DIAGNOSIS: - ECTODERMAL DYSPLASIA

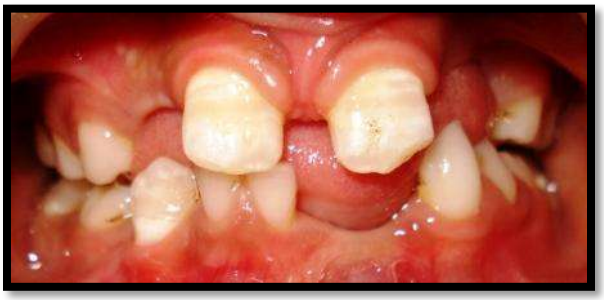

Fig 3: Intra-oral view showing hypodontia

INVESTIGATIONS

Radiographic: Orthopantomogram was taken which revealed anodontia of certain teeth **(12 13 22 23 24 31 32 41 42 43)** as marked in the Figure 4.

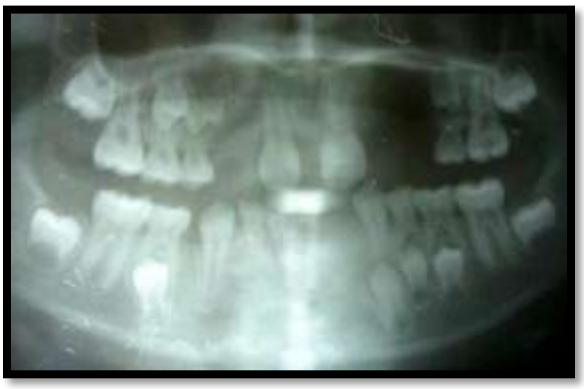

Fig 4: A panoramic radiograph revealing anodontia of 10 teeth

Sweat pore count (Table1):

The sweat pore count in 1 cm2 on the palm of the patient was 6 which were on the lower side of normal count.

TABLE 1

	Number/cm2
PATIENT	6
NORMAL VALUE	12-14

DIFFERENTIAL DIAGNOSIS:-

- DOWN'S SYNDROME
- GOLDENHER SNYDROME
- CROUZON SYNDROME
- CLOUSTON SYNDROME

DIAGNOSIS: - CST SYNDROME

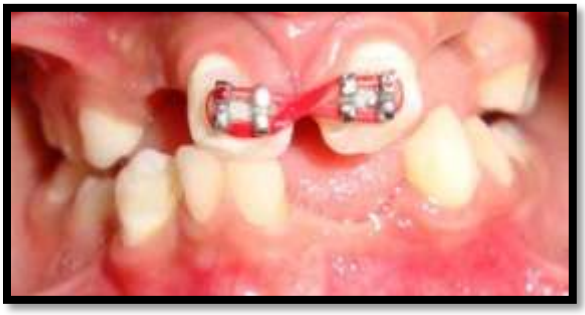

Fig 5: Showing midline diastema

MANAGEMENT:

- A multidisciplinary approach was performed-
  - Oral prophylaxis and application of topical fluoride.
  - Restoration of left maxillary central incisor with composite.[Figure 5]
  - Closure of the midline diastema using orthodontic brackets and elastics. [Figure 6&7]

- Cast partial denture in relation to **12 13 22 23 24** and removable partial denture in relation to **31 32**.[Figure 8&9]
- Recall checkup after every 3 months. [Figure 10]

Fig 6: Composite resin build up for left maxillary central incisor

DISCUSSION

Ectodermal dysplasia often involves overlapping features, thereby complicating a definitive classification. Diagnosis becomes difficult as the characteristic features are not obvious during birth, though during neonatal period there is extensive scaling of the skin and unexplained pyrexia (Guckes et al, 1991). After diagnosis, the parents of the affected patient can be counseled regarding the treatment options available. (1)

Fig 7: Closure of midline diastema using MBT 0.002 inch bracket

Literature shows children rejected by their peer groups are more likely to become aggressive, delinquent, and may experience mental health problems in adulthood. Therefore, successful treatment of the present case can be expected to assist the patient both physically and psychologically. Clinical reports have stated the importance of prosthetic dental treatment in patients with anodontia or hypodontia for physiological and psychosocial reasons. (3) The successful use of any prosthesis is dependent on the cooperation and communication between the dental team and the patient and his parents. [6,7] Diagnosis was not just based on clinical triad but also on family history, radiographic findings and sweat pore count which were all suggestive of CST syndrome.

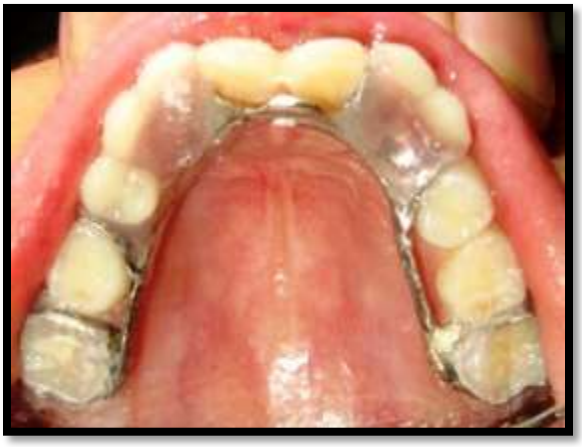

Fig 8: Cast partial denture in relation to 12 13 22 23 24

Treatment of a child with ectodermal dysplasia requires a multidisciplinary approach which generally includes a removable and /or fixed partial denture, an overdenture, complete denture prosthesis or an implant retained prosthesis. In cases where there is associated cleft lip and palate the treatment may consist of intervention by a plastic surgeon and an oral and maxillofacial surgeon.

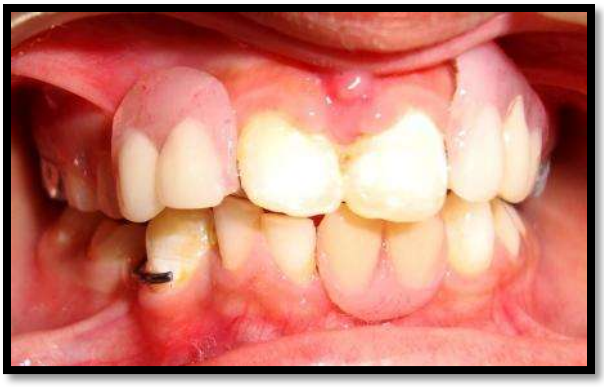

Fig 9: Removable partial denture in relation to 31 32

These treatment modalities can be used individually or in combination to provide an optimal result. The proper sequencing of treatment is important to achieve the desired function and aesthetic results.[3,8,9,10] In the present case, closure of the midline diastema was carried out using orthodontic brackets and elastics and the left maxillary central incisor was restored using composite. This was followed by prosthodontic management by cast partial denure in maxillary arch and removable partial denture in mandibular arch. As the child matures the removable prosthesis needs relining, rebasing or remaking to accommodate growth changes and maintain normal oral functions. Due to the girl's young age, on-going development of the jaws and insufficient quantity of alveolar bone, endosseous implant placements were not possible. When child reaches teenage years, orthodontic treatment may be indicated as better management of spacing may prepare the mouth for a fixed partial denture or implants in future.

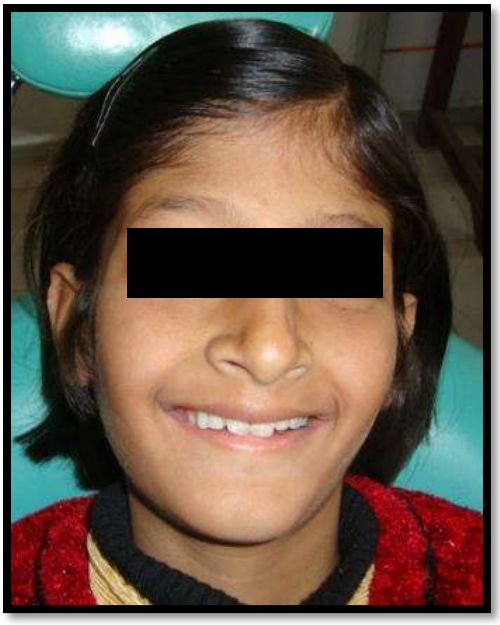

Fig 10: Post- operative patient's photograph

CONCLUSION

Young patients with ectodermal dysplasia need to be evaluated early by a dental professional to determine the oral ramifications of the condition. This clinical report describes the types, characteristic features and treatment options for a young female patient with ectodermal dysplasia. The unaesthetic appearance, poor self image, school/job related discrimination often accompanies ectodermal dysplasia syndrome which has a negative psychological effect on the patient. Thus management of the orofacial disfigurement not only provides dental benefits but also provides some measure of confidence as well as psychological benefits. Therefore, appropriate care needs to be rendered throughout the child's growth cycle to maintain oral functions as well as to address the esthetic needs of the patient.

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