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PROSTHETIC MANAGEMENT OF CHRIST-SIEMENS- TOURINE SYNDROME – A CASE REPORT

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ABSTRACT: Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. The X-linked recessive ED (Christ-Siemens-Touraine syndrome) is the most common disordered (80% of EDs); it affects males and is inherited through female carriers. It is characterized by the triad of signs comprising of sparse hair (atrachosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The lack of teeth and the special appearance were reported to be major concerns. The incidence in male is estimated at 1 in 100,000 births, the carriers-incidence is probably around 17.3 in 100,000 women. Early dental intervention can improve patient's appearance, thereby minimizing the associated emotional and psychological problem in these patients. Treatment requires team work by medical personal along with dental professionals. This clinical report describes the prosthetic rehabilitation of 24-year-old male patient with multiple missing and malformed teeth along with other classical oral and general manifestation of Christ-Siemens-Touraine syndrome.

KEYWORDS: Ectodermal Dysplasia, Christ-Siemens-Touraine syndrome, hypodontia, hypotrichosis, hypohidrosis.

INTRODUCTION: The ectodermal dysplasias (EDs) comprise of a large, heterogeneous group of inherited disorders that are by primary defects in the development of 2 or more tissues derived from embryonic ectoderm. The tissues primarily involved are the skin, hair, nails, endocrine glands, and teeth. The syndrome involves overlapping features, thereby complicating a definitive classification. Lamartine[1] in 2003 has described various well defined ectodermal dysplasia as Hypohidrotic (anhidrotic), Hidrotic (Clouston's syndrome), Ectrodactyly-Ectodermal dysplasia-cleft syndrome (EEC), Rapp- Hodgkin Syndrome, Hay-Wells syndrome or ankyloblepharon ectodermal dysplasia. Viera et al in 2007[2] classify ectodermal dysplasia based on the number and function of sweat glands involved as mentioned below:

Hypohidrotic (anhidrotic) Ectodermal Dysplasia (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. 60-70% of cases usually show general manifestations as 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 characterised reduction in amount of hair (hypotrichosis), absence of sweat glands (anhidrosis) resulting in temperature elevation, absence of sebaceous

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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.[2,3]

The oral manifestations include conical or peg shaped teeth, hypodontia or complete anodontia of both the deciduous and the permanent dentition with malformation of any teeth that may be present, generalized spacing, underdeveloped alveolar ridges and delayed eruption of permanent teeth. Even when complete anodontia exists the growth of the jaw is not impaired. This would imply that the development of the jaws except for the alveolar process is not dependent upon the presence of teeth. However, since the alveolar process does not develop in the absence of teeth, there is a reduction in the normal vertical dimension resulting in the protuberant lips. In addition, in oropharynx the defect may be manifested as a high palatal arch or even a cleft palate. The salivary glands including the intraoral accessory glands are sometimes appear hypoplastic in this disease. This result in xerostomia and the protuberant lips may be dry and cracked. [2-5]

Hidrotic Ectodermal Dysplasia (Clouston Syndrome): The clinical features include nail dystrophy, hair defects and palmoplantar dyskeratosis. The patients have normal facies, normal sweating and no specific defect is seen. [4]

CASE REPORT: A twenty four year old male patient, reported to the Department of Prosthodontic, Sharad Pawar Dental College, Sawangi, Wardha with a complaint of missing teeth in upper and lower jaws. No history of exfoliation of teeth but history of delayed eruption of teeth. Patient also complained of dry skin and absence of sweat in his skin as a result of which he is not able to withstand hot water and hot environment. Patient gives history of dry mouth and difficulty in swallowing. Extra oral examination reveals patient has less hair in eyebrows and eyelashes. There was prominent frontal bossing, depressed nasal bridge and protuberant lips (Fig 1&2). Skin of face, upper and lower limbs was dry, with normal nails and scanty eyebrows. Family history was not contributory. Intraoral examination revealed patient had partially edentulous upper and lower arches. Teeth present in maxillary arch were 11, 16, 17, 21, 26, 27 and 36, 46 in mandibular arch (Fig 3). After correlating the finding the patient was diagnosed of Hypohidrotic (Anhidrotic) Ectodermal Dysplasia. Treatment planning for the case included over denture for maxillary arch with coping on upper central incisor. Intentional root canal treatment was carried out on upper 11, 21, followed by metal coping. A silicone impression material was used for impression making with dry mucosa, because it is hydrophobic and requires an extremely dry field [6]. Jaw relation record were made and anatomic 33-degree, acrylic posterior teeth were used to develop bilateral-balanced occlusion. Try in of maxillary and mandibular denture to verify centric relation and VDO was carried out. After the approval of the tooth arrangement by the patient, the wax up dentures were processed in a heat-polymerized denture base resin (Lucitone 199, Dentsply International Inc, York, Pa.). Fit of the completed dentures was verified (Fig. 4) with a pressure-indicator paste (Mizzy Inc, Cherry Hill, N.J.) and disclosing wax. Follow up appointments were carried out regularly.

DISCUSSION: Ectodermal Dysplasia often involves overlapping features, thereby complicating a definitive classification. Some Ectodermal Dysplasia Syndrome may be mild whereas others may be devastating. Diagnosis becomes difficult as the characteristic features are not obvious during birth,

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though during neonatal period there is extensive scaling of the skin and unexplained pyrexia [1, 2, 7]. After diagnosis, the parents of the affected patient can be counseled regarding the treatment options available. Preservation of alveolar bone is imperative in individuals with ED; because they must depend on the alveolar ridges for prosthesis support from an early age [4-8].

Mutations in the gene Ectodysplasin-A receptor (EDAR) and Ectodysplasin-A Receptor-Associated Adapter (EDARADD) are also responsible for the rare autosomal dominant and autosomal recessive forms [9, 10]. The X-linked recessive HED has full expression in males. Female carriers are more than affected men, but they have little or no signs of the condition. Zhang et al [11] have described some correlations between the phenotypes and genotypes of X-linked HED and nonsyndromic hypodontia subjects. They suggested that nonsyndromic hypodontia is probably a variable expression of X-linked HED. Significant differences in the number of primary missing teeth have been detected between X-Linked HED and autosomal recessive HED [12].

Periodic recall of young ED patients plays important role because prosthesis modification or replacement will be needed as a result of continuing growth and development. In addition to adjustments related to fit, the occlusion of prosthesis must be monitored for changes because of jaw growth. [13] Other problems related to removable prostheses are speech difficulties, dietary limitations, and loss of the prosthesis [4-8]. Endosseous implants can also be considered as an alternative treatment. [14, 15]

SUMMARY: This clinical report describes the characteristics and prosthodontic restoration of a young male patient Christ-Siemens- Tourine Syndrome. With proper care and prosthodontic treatment, the patient can enjoy a relatively normal life. It is important that the patient and his parents fully understand the dental problems related to his physiologic and psychologic conditions. The need for continued dental treatment is necessary. Recall visits plays an impotent role in rehabilitation of these patients.

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FIGURES:

Fig 1&2: Extra oral view showing Frontal bossing and depressed nasal bridge, protuberant lips.

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Fig 3: Pretreatment intra oral view

Fig 4. Post treatment intra oral view

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