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Christ-Siemens-Touraine syndrome – A report of a rare case and a multidisciplinary treatment plan with literature review

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KEYWORDS

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ABSTRACT:

Aberrant development of ectodermal structures and congenital dysplasia of one or more accessory appendages is a characteristic of a heterogeneous group of disorders known as ectodermal dysplasias [EDs]. These disorders are widespread, nonprogressive, and congenital. Education, counselling, prosthetics, and general rehabilitation are the main focuses of a multidisciplinary approach to the treatment of patients with ectodermal dysplasias and their families. The article presented here highlights a rare case of Christ-Siemens-Touraine syndrome in a 15-year-old male patient with a multidisciplinary treatment plan.

1. Introduction

Ectodermal dysplasia (ED) is a diverse set of inherited disorders that are characterised by shared structural and functional abnormalities in ectoderm-derived tissues such as the sweat glands, teeth, hair, or nails. [1] Based on the pathways involved, ectodermal dysplasia is presently classified into four categories of disorders: the wingless type MMTV integration site family, member 10 [wnt 10], tumour protein p63, the ectodysplasin/nuclear factor-kappa B [NFkB] pathway, and the structural group.[2]

One category of hypohidrotic ectodermal dysplasia (HED) is Christ-Siemens-Touraine syndrome. Defects in the ectodysplasin signalling system, which is essential for the development of ectodermal appendages during embryonic development, cause the illness.

Despite efforts to separate the different disorders, there is a significant amount of clinical feature overlap, which makes a comprehensive history and clinical examination crucial in assisting us in making a diagnosis and evaluating the various systems involved at play.[2]

Christ-Siemens-Touraine syndrome has X-linked mode of inheritance and is characterized by hypohidrosis, hypotrichosis and hypodontia.[3] About 1 in 100,000 liveborn males have the X-linked variant of hypohidrotic ectodermal dysplasia[HED], which affects people of all races and ethnicity. Although female carriers may have dermatoglyphic anomalies, sparse hair, reduced sweating, and dental abnormalities, the entire condition does not occur in females.[4]

Here we report a rare case of Christ-Siemens-Touraine syndrome in a 15 years old male patient with multidisciplinary management plan and literature review.

2. Case Report

A 15-year-old male patient visited the Department of Pediatric and Preventive Dentistry with a chief complaint of missing teeth in his oral cavity during early childhood and a feeling of dryness in his mouth. Family history was contributory- his mother and younger brother was also affected and there was no history of complications during his delivery. Past dental history was nothing significant. On extraoral examination, the patient had typical facies characterized by prominent supraorbital ridges, saddle nose, thick everted lips, midface hypoplasia with low ears, and sparse eyebrows. Upon inspection, the hair seemed very fine sparse with lustreless appearance. Eye examination showed dryness of the cornea and decreased lubrication/ tearing **[Figure 1]**. Examination of the fingers showed thin, fragile nails and normal-shaped fingers. Upon skin inspection, the patient had dry, scaly skin and a slightly elevated fever .

On intraoral examination the patient showed oligodontia with missing of 11, 12, 14, 17, 21, 22, 24, 27,31, 32, 33,

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34, 35, 36, 37, 41, 42, 43, 44, 45, 46, 47 with over retained deciduous teeth wrt 53,55, 63,65 was seen **[Figure 2]**. Teeth present in the anterior region of maxilla were conical. Vertical heights of both maxillary and mandibular alveolar ridges were less.

On radiographic examination, orthopantomogram[OPG] revealed deciduous teeth wrt 53,55,63,65 and permanent teeth wrt 11, 15,16 and 26. The patient had an undeveloped maxillary and mandibular alveolar ridge. No evidence of secondary teeth germs was evident. **[Figure 3]**.

Figure 1: Extraoral photograph showing prominent supraorbital ridges, saddle nose, thick everted lips, midface hypoplasia with low ears, sparse eyebrows and hairs.

Figure 2: A) Oligodontia with the presence of 53,55,16,63,65,26 in the maxillary arch.

B) Edentulous mandibular arch.

Figure 3: Orthopantomogram showing the presence of 11,53, 55, 15,16, 63, 65, 26. Undeveloped maxillary and mandibular ridges. No evidence of secondary teeth germs was evident.

Treatment plan: Till the age of 19 years, the preferred treatment option in this case is a single complete denture for the mandibular arch, orthodontic extrusion of 11, followed by removable partial denture wrt 12,14,21,22 and 24. At the age of 19 years, the preferred treatment is ridge augmentation followed by implant placement using All-on-4 concept in mandible and single tooth implant placement wrt 12,14,21,22 and 24.

3. Discussion

Genetic defects in the ectodysplasin signal transduction system is the cause of Christ-Siemens-Touraine syndrome.[3] In this case, the ectodysplasin/nuclear factor-kappa B [NFkB] pathway is affected. Multidisciplinary treatment is essential for individuals with Christ-Siemens-Touraine syndrome since there are several clinical and psychological factors that has to be addressed. Restoring oral function and avoiding hyperthermia are the main goals of therapy.[5]

Early prosthodontic intervention aids in the child's adjustment to the prosthesis and promotes proper temporomandibular joint function, speech, mastication, swallowing, and aesthetic appeal. During times of rapid

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growth or growth spurts, the intraoral prosthesis can be adjusted. An early age intervention has positive psychological effects in addition to dental ones.[6] In our case, we have planned a single complete denture for the mandibular arch and removable dentures for the maxillary arch till 18 years of age because of intense and ongoing skeletal growth and development of the orofacial system. Bone growth and density often incomplete at this age. Placing dental implants can interfere with jaw growth and dental development.

Prosthodontic rehabilitation is difficult because of the reduced alveolar bone height, "knife-edge" morphology, and the limited amount of tooth structure that remains. Implants are often only used to replace teeth in children whose craniofacial growth is complete, and it is probably preferable to wait until puberty. Implant failure is more likely among ED patients under the age of 18. Implant placement in children or teenagers may result in a number of undesirable side effects, such as multifaceted restrictions on skeletal craniofacial growth and damage to tooth germs. Because of continued growth and development as well as inadequate alveolar bone support, implant therapy was not the best course of action in this instance. The dentures should be regularly evaluated by the pediatric dentist as per the child's growth and development and are modified/replaced accordingly [7].

In addition to replacing missing teeth and providing instant cosmetic effects, flexible dentures also enable patients to become used to removable prostheses prior to receiving final prostheses. Because of its advantageous qualities, such as strength, biocompatibility, patient comfort, and aesthetics, a Valplast flexible partial denture was planned for the maxillary arch [8].

Preventive measures for hyperthermia include avoiding hot environments and strenuous physical activities, using air conditioning, cooling vents and for maintaining proper hydration and thermoregulation, it would be prudent to consume cold drinks often. Artificial tears and saliva may be beneficial for patients with decreased lacrimation and xerostomia, respectively.[5] Topical minoxidil has been used to treat alopecia, with some individuals showing positive outcomes.[2] Use of wigs is advised for people with severe alopecia. If abnormalities in word-articulation and phonetics are found, consultation with an otolaryngologist and a speech therapist is indicated. Genetic counselling for family planning and Regular psychological counselling are essential for individuals and families affected by ectodermal dysplasia.[5]

Table 1 Summarizes the literature regarding various dental treatment options for Ectodermal Dysplasia depending on the clinical scenario.

Author	Year	Age/sex	Clinical findings	Treatment done
Hekmatfar S et al [9]	2012	10yrs/F	Presence of 11,55,16,21,65,26, 75,36,37,41, 85 and 46	Crossbite was corrected with a Hawley appliance and a Removable partial denture for the replacement of missing teeth.
Khan M W et al [10]	2019	19yrs/M	Presence of 11,13,21,23 and 43	Single visit Intentional root canal treatment of all the permanent teeth was done and removable complete overdentures was given for both the arches.
Machado M F et al [11]	2022	16yrs/M	Presence of 11,55,21 and 65	Initially treated with a removable partial denture for maxillary arch and single complete denture for mandibular arch. A combination of autogenous and allogenic bone grafts procedure was performed under local anesthesia when the patient was 19 years old. Implant supported prosthesis was placed when the patient was 21 years old.
Zhu Q et al [12]	2021	19yrs/M	Presence of 65, 26, 36, 37, 85, and 46,	Initial surgery: maxillary augmentation and right maxillary sinus external lift. Second surgery (16 weeks after first surgery): mandibular augmentation Third surgery (40 weeks after first surgery): maxillary implant placement and apically repositioned flap. Fourth surgery (50 weeks after first surgery): mandibular implant placement
Elgasmi FE et al [13]	2025	10yrs/F	Presence of 12,55,16,22,65,26,3 2,75,36,85 and 46	Removable partial denture for both maxillary and mandibular arches for the replacement of missing teeth.
	2025	15yrs/M	Presence of 53,55,16,63,65 and 26	Removable partial denture for maxillary arch and single complete denture for mandibular arch. At the age of 19yrs ridge augmentation followed by implant placement using All on 4 concept in mandibular arch and single implants are planned for 12,14,21,22 and 24

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4. Conclusion

Rapid diagnosis and prosthetic rehabilitation using a multidisciplinary approach are critical to the success of ED care. Pedodontists play a vital role in managing ED because they are highly trained in child psychological and behavioural treatment.

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