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Christ-Siemens-Touraine Syndrome: Report of a case with mini review

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Abstract

Christ-Siemens-Touraine syndrome, commonly known as Hypohidrotic Ectodermal Dysplasia is the best studied variant of Ectodermal Dysplasia which is a spectrum of a large group of inherited developmental syndromes. This syndrome is primarily characterized by a triad of hypodontia, hypotrichosis and hypohidrosis, with or without a defect in other ectodermal structures such as ears, lips, digits, dermatoglyphics. However, the oral manifestation of hypodontia or occasionally complete anodontia is a challenge to one's self-esteem. Therefore, oral rehabilitation through a multidisciplinary approach becomes an important concern in such cases. Here, a case of Hypohidrotic Ectodermal Dysplasia in a 22 year-old male has been reported. Patient was treated with removable characterized over-denture with age appropriate artificial teeth, which significantly boosted his self-esteem and improved speech, masticatory function and facial aesthetics.

Keywords: Ectodermal Dysplasia, hypodontia, hypohidrosis, hypotrichosis

Introduction

Ectodermal Dysplasia (ED) syndrome is a rare hereditary disorder of ectodermal origin. This syndrome has been well documented in the dental literature, which shows an incidence of about 7 per 10,000 live births.[1] Thurnam in 1848,[2] first reported of this syndrome but the term ED was coined by Weech in 1929.[3] Freire-Maia and Pinheiro[4] proposed the first classification system of ED with subsequent updates. They classified the patients into subgroups which were based on the presence or absence of four primary defects including Trichodysplasia (abnormal hair), Dental dysplasia (abnormal dentition), Onychodysplasia (abnormal nails) and Dyshidrosis (abnormal or missing sweat glands), with or without a defect in other ectodermal structures. To date, more than 192 distinct disorders have been described, the most frequently reported phenotype being Hypohidrotic ED (Christ-Siemens-Touraine syndrome).[5] Presented here is a case report of a 26 year old male patient with hypohidrotic ED.

Case Report

A 26-year-old male reported with a chief complaint of missing teeth in upper and lower jaws since childhood and desired artificial replacement of his missing teeth. Patient gave no history of exfoliation or extraction of teeth but gave a history of delayed tooth eruption. He further complained of dryness of skin, intolerance to heat, dry mouth and lack of sweating. When enquired about a similar history in the family, patient gave a positive history of his maternal grandfather suffering from a similar

complaint and further reported of consanguineous marriage of his apparently healthy parents.

On extra oral examination (Figure 1), patient had fine sparse lustreless hair over the scalp, scanty eyebrows and eyelashes, prominent supraorbital ridges, saddle nose, sunken cheeks, thick everted protuberant lips and prominent obliquely set ears. However, nails appeared normal. Intraoral examination (Figure 2) revealed cone-shaped maxillary centrals with midline diastema, high arched palatal vault, presence of 16 and 26, presence of retained 55 and 65 and an underdeveloped completely edentulous mandibular alveolar ridge. Thus, based on the history, extraoral and intraoral clinical findings, a diagnosis of Hypohydrotic ED was made. An Orthopantomogram (Figure 3) confirmed the clinical findings.

Treatment plan which included fabrication of characterized complete over-denture for the maxillary arch and a conventional complete denture for the mandibular arch, was executed by the department of prosthetic dentistry (Figure 4).

Discussion

Hypohidrotic ED is a rare congenital disorder inherited as X-linked (most common), autosomal dominant or autosomal recessive trait, the probable aetiology being pathogenic mutations in the genes ectodysplasin A (EDA), ectodysplasin A receptor (EDAR) or ectodysplasin A receptor- associated death domain protein respectively, which alter various signalling pathways involved in ectoderm-mesoderm interactions, differentiation of ectodermal appendages and organogenesis during the initiation of embryonic development.[6]

In the present case, as evident from clinical history, the patient and his maternal grandfather both were affected, ascertaining the mode of inheritance to be X-linked recessive.

X-linked Hypohidrotic ED shows full expression only in males while females are the carriers. Though the condition is clinically characterized by the classical triad of hypotrichosis, hypodontia and hypohidrosis, a plethora of extraoral and intraoral manifestations have been documented in the literature. The extraoral features include fine sparse lustreless hair over the scalp, scanty eyebrows and eyelashes, frontal bossing, sunken cheeks, saddle nose, thick everted protuberant lips, wrinkled hyper pigmented periorbital skin, prominent supraorbital ridges, prominent obliquely set ears, extensive scaling of the skin and occasional nail deformities. A typically depressed mid-face and a collapsed lower anterior facial height imparts a typical aged appearance to the face. Furthermore, patients may have no or decreased sweating because of lack of sweat glands, producing pyrexia as their skin cannot control body temperature properly. Asthma, eczema, chronic nasal infections with foul smelling discharge and increased lung infections are occasionally reported in these patients. Intraorally, both the deciduous and permanent dentitions are affected. The dental characteristics include peg-shaped or conical front teeth, hypoplastic teeth, hypodontia (more common) or complete anodontia and underdevelopment of the alveolar ridges when the teeth are missing. [5, 7]

The presentation of extraoral and intraoral characteristics observed in the present case is in absolute agreement with the existing literature.

In patients with ED, diagnosis is done based on family history, thorough clinical and radiological examinations and the management mandates a multidisciplinary approach, as follows:

1. Since the affected individuals suffer from low self-esteem due to their unusual physical appearance and lack of social acceptance, psychological counselling should be advised on a regular basis.
2. Consultation with a speech therapist and an otolaryngologist is warranted if difficulty in speech articulation is detected.
3. Patients are advised to use wigs in case of severe alopecia. [8]
4. Dental management should be provided to improve the cosmetic appearance of the patient. The treatment modalities for prosthetic rehabilitation of such patients include fabrication of complete dentures, removable partial dentures, over-dentures or implant retained prostheses. However, treatment of each patient should be customized according to the patient's age, growth and development and available bone support. [9]

In the present case, characterized complete over-denture is preferred over conventional removable prostheses which require frequent adjustments to improve compromised denture retention and stability

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and over implant retained prosthesis which demands presence of sufficient alveolar bone height and width which is deficient in the present case. On the contrary, over-denture preserves alveolar bone, preserves proprioception, preserves existing natural dentition and improves retention and support without much of a prerequisite. [9]

As a future prospective, though still in a trial phase, intravenous or intra-amniotic injections of recombinant EDA-A1 might alleviate some of the symptoms of X-linked Hypohidrotic ED as these have been found to restore the growth of teeth, skin structures and mucous glands in laboratory animals.[10]

Conclusion

Ectodermal dysplasia affecting young individual presents a unique challenge in terms of management of its clinical manifestations which contribute to oral malfunction, defective phonetics and unappealing facial aesthesis leading to considerable social and psychological set back in these patients. Therefore, a timely correct diagnosis with relatively prompt, easy, acceptable and economical solution is desirable to boost up patient's self-esteem and optimize his social integration.

Referrences

1. Tarjan I, Gabris K, Rozsa N. Early prosthetic treatment of patients with ectodermal dysplasia: a clinical report. Journal of Prosthetic Dentistry 2005; 93(5): 419–24.
2. Thurnam J. Two cases in which the skin, hair and teeth were very imperfectly developed. Proc RM Chir Soc 1848; 31: 71-82.
3. Weech AA. Hereditary ectodermal dysplasia. (Congenital ectodermal defect). Am J Dis Child 1929; 37: 766-90.
4. Pinheiro M, Freire-Maia N. The ectodermal dysplasias. Arch Dermatol 1982; 118(4):215-6.
5. Shah KN. Ectodermal Dysplasia. eMedicine Article last updated: Feb 11, 2019.
6. Trzeciak WH, Koczorowski R. Molecular basis of hypohidrotic ectodermal dysplasia: An update. J Appl Genet 2016;57: 51-61.
7. Vierucci S, Baccetti T, Tollaro I. Dental and craniofacial findings in hypohidrotic ectodermal dysplasia during the primary dentition phase. The Journal of Clinical Pediatric Dentistry 1994; 18(4): 291-7.
8. Meshram GG, Kaur N, Hura KS. A case report of hypohidrotic ectodermal dysplasia: A mini-review with latest updates. J Family Med Prim Care 2018; 7: 264-6.
9. Thayer HH, Caputa AA. Effects of over-dentures upon remaining oral structures. J Prosthet Dent 1977; 42: 374-81.
10. Hermes K, Schneider P, Krieg P, Dang A, Huttner K, Schneider H, et al. Prenatal therapy in developmental disorders: Drug targeting via intra-amniotic injection to treat X-linked hypohidrotic ectodermal dysplasia. J Invest Dermatol 2014; 134: 2985-7.

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Figure 1: A. Pre-operative frontal view

B. Pre-operative lateral view

C. Dry skin and absence of nail deformity

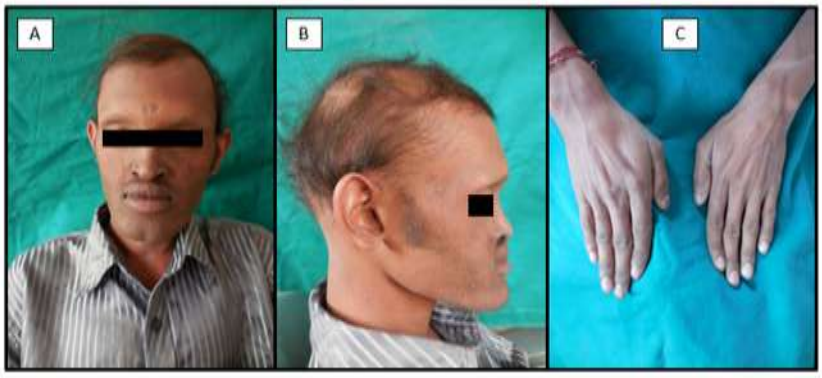


Figure 2: A, B, C. Pre-operative intraoral examination

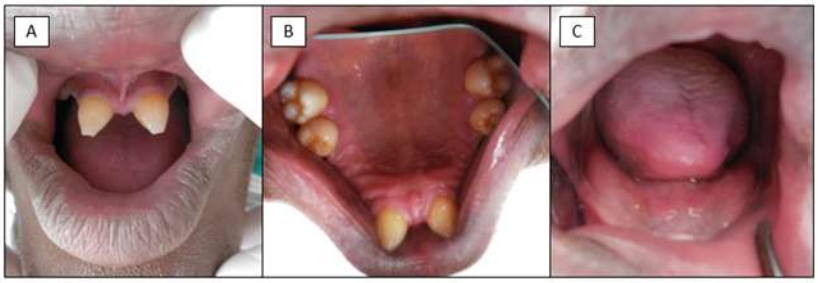
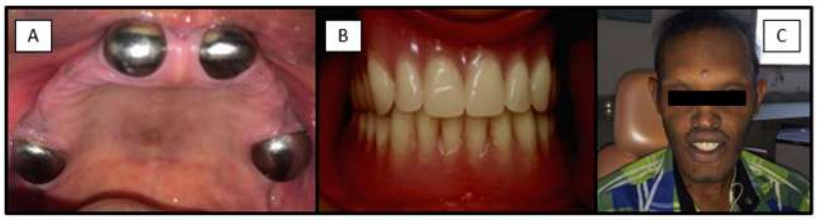


Figure 3: A. Metal copings

B. Final denture

C. Post-operative frontal view



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