

Presentation and management of ectodermal dysplasia: case report

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Abstract

Ectodermal dysplasia is a clinically and genetically heterogeneous group of disorder with about 192 rare traits having absent, incomplete or delayed development of one or more of the four ectodermal appendages of hair, teeth, nails and sweat glands. Oral manifestations include agenesis of the primary and permanent teeth, which manifest clinically as hypodontia, or anodontia. A case of ectodermal dysplasia is reported with clinical presentation of prominent lips, nasal bridge and forehead, wrinkling and hyperpigmentation of peri-orbital and peri-oral region. The patient also presented with hypodontia and conical incisors. Restoration of malformed teeth was carried out and missing teeth were replaced with removable prosthesis. Successful dental treatment involves a holistic approach with input from the various dental specialties.

Key words: Ectodermal dysplasia, Hypodontia, Restoration, Prosthesis

Introduction

Ectodermal dysplasia is a clinically and genetically heterogeneous group of disorder with about 192 rare trait having absent, incomplete or delayed development of one or more of the four ectodermal appendages of hair, teeth, nails and sweat glands^{1,2}.

There are a number of classifications for this condition. However, the current classification system is based on clinical manifestations that assign the syndrome numerically, depending on the number of appendages involved-hair, teeth, nail, and sweat glands dysplasia^{3,4}.

Hypohidrotic X-Linked ectodermal dysplasia is the most common ectodermal dysplasia with a birth frequency of 1:10,000 to 1:100,000⁵. The aetiology of X linked heterogeneous ectodermal dysplasia has been associated with mutations in the ectodysplasin-A1 gene which codes for the tumour necrosis factor family member ectodysplasin-A (EDA)^{6,7}.

The clinical features commonly include sparse, dry, fine and short hair. Hair on eyelashes and eyebrow may be sparse or absent. The skin may be smooth, dry, and finely wrinkled especially around the eyes and those affected may age prematurely. The nails are abnormal in 50% of patients, and appear brittle, thin or ridged. There is reduced ability to sweat, as the number of sweat glands is reduced⁸.

Oral manifestations are that of agenesis of most of the primary and permanent teeth, which manifest clinically as hypodontia or anodontia. Other oral characteristics include impacted teeth, altered tooth morphology (such

as peg-shaped or conical teeth) and reduced saliva production^{9,10}. There is also decreased facial height due to lack of teeth, protruding lips and under developed alveolar ridge¹¹. Symptoms are milder and vary in female carriers, due to the X-chromosome inactivation^{1,8}.

The teeth, when present, may be hypoplastic and often there is xerostomia as a result of salivary gland hypoplasia. There may also be gingival disorders. There is associated heat intolerance and discomfort. Hyperpyrexia may be the reason for medical emergency especially in children. There is also a characteristic facies, everted lips and large ears. Growth may be stunted with associated mental retardation¹¹.

Dental treatment is often lengthy and complicated, with treatment initiated by the paediatric dentist at an early age (from the second year)^{12,13}. Other treatment, such as provision of prostheses are instituted in later years. These prostheses include partial dentures, full dentures or fixed prosthetic treatment¹⁴.

The aim of this paper is to report a case of ectodermal dysplasia managed in tertiary centre and the outcome.

Case Report

A 14-year-old male presented with a history of non-eruption of most of his teeth. The child, according to the father, had to withdraw from school because of the embarrassing condition. On enquiry, the patient claimed occurrence was observed in the grandfather.

Physical examination revealed a characteristic old man facies (prominent lips, nasal bridge and forehead, wrinkling and hyperpigmentation of perioral and periorbital small sized nose with nostril hypoplasia). The hair on eyelashes was scanty and there was no eyebrow (Figure 1). The skin all over the body was thin and wrinkled. Nails were however normal. Intraoral examination showed erupted maxillary centre and lateral incisors which were conical in shape and atrophic lower alveolar ridge (Figure 2). Systemic examination showed no abnormality. The patient's orthopantomograph showed erupted conical upper left and right lateral incisors and canines and no other teeth.

A provisional diagnosis of anhidrotic hereditary ectodermal dysplasia was made based on the clinical features.

Treatment

Treatment protocol included appointment with parents and patient in order to establish rapport, enhance acquaintance and confidence in the dentists.

Evaluation of treatment was done and the decision for restoration of conical incisors to normal morphology using composite restoration was made. Fabrication of removable partial denture for the upper jaw and complete dentures for the lower was also planned.

The pediatric dentists carried out composite build up of the conical teeth.

Fabrication of removable dentures was done by the prosthodontist. An alginate impression was taken using a metallic impression stock tray. The impression was casted in dental stone. A special tray was fabricated in cold cure acrylic; this was used to make secondary impression. Green stick impression compound was used for border moulding while light body addition silicone impression material was used for the wash impression. The impression was casted in dental stone. Bite registration was done and later proceeded to the try-in of the trial denture. The definitive prostheses were then fabricated following patient and parents' expressed satisfaction with the trial denture.

The dentures were fitted and mucous membrane lubricant was given to enhance easy wear of denture and comfortability (Figure 3): Patient was followed up for a year; patient wore the denture but did not go for management of his skin problems and counseling. The father was advised on the importance of proper management.

Outcome

The treatment was successful, with satisfaction of the patient, his parents, as well as the dentists. The patient kept appointment for adjustment visits and had to learn to adapt to denture wearing since this was his first denture. He gradually gained good retention and stability over the first year. He was also able to adapt because he was eager to wear the denture to stop the teasing by his peers.



Figure 1: The facial characteristics of the patient at presentation.



Figure 2: Conical upper incisors with atrophic lower alveolar ridge



Figure 3: Post operative appearance of the patient

Discussion

Patients with ectodermal dysplasia exhibit a variety of clinical characteristics with a typical triad: hypohidrosis, hypodontia/oligodontia and hypotrichosis. The exhibited features of this condition are the following manifestations: salivary and sweat glands alterations, scaling of the skin, breathing difficulties, hearing loss, wrinkles around the mouth and orbits, peri-orbital hyperpigmentation, prominent lips, narrow, hypoplastic maxilla and heat intolerance. The males have an easily recognisable facies, also known as an "old man"¹⁵. These features are similar to those described in previous reports¹⁶⁻¹⁹. The missing teeth experienced by the patient in this

report was also a source of worry to the parents²⁰. For the young patient, there is a delay in teething which lead to the diagnosis in the second year of life²⁰. Hypodontia may affect both primary and permanent dentitions with variable number of missing teeth¹⁵.

The experience with the male patient in this report further supports the finding in previous studies that dental treatment should be instituted as early as possible to avoid teasing and social isolation^{21,22} and thus improved quality of life. It has been reported that merely looking different from one's peers may result in lowered self esteem, speech defects, decreased academic performance and social isolation²³. Therefore, the child's psychological health should be given important consideration. It is suggested that provision of partial or complete denture should be as early as two to three years and dentures modified periodically because of changes in alveolar growth, erupting teeth, occlusal and basal dimensions²⁴. Dentures have documented drawbacks and these include loss of prostheses, limited retention and stability, accelerated alveolar bone resorption of existing hypoplastic process and restriction of growth at the mid-palatal suture. Restoration with crowns and bridges have also been suggested in children¹⁵, but the use of dental implant is not advised until cessation of growth²⁴. The documented success of implant placement in edentulous ectodermal dysplasia patient is variable²⁵. Parent's cooperation is also necessary to ensure success of treatment as reported in a previous study²². In the management of these patients, it is essential to evaluate and counsel before treatment. This is because affected families have different ways of managing the challenges that arise when a family member has a genetic condition²⁶.

The patient had composite restorations of the conical teeth with fabrication of prosthesis. Restoration of existing teeth can be modified with direct or indirect composite crowns. This improves aesthetics²⁴.

The treatment provided a significant improvement in esthetics, masticatory and phonetic function. There was also an improvement in the child's self esteem.

Research has shown that the administration of recombinant EDA-A1 may provide possible treatment in ectodermal dysplasia²⁷. Experimental studies show that the recombinant EDA-A1 may correct the pathological features of linked hypohidrotic ectodermal dysplasia²⁸. Treatment of ectodermal dysplasia is directed towards specific symptoms that are apparent in each individual. The optimal treatment of these patients should involve paediatric dentists, orthodontists, prosthodontists, dermatologists and a clinical geneticist²⁴. Presently, there is no structured multidisciplinary approach to the management of ectodermal dysplasia exist in Nigeria.

Conclusion

The successful management of patient with ectodermal dysplasia involves the input of various specialties in dentistry. It is however important that the several recall visits and lengthy procedure be explained to patients and parents.

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