A CASE REPORT OF HYPOHIDROTIC ECTODERMAL DYSPLASIA IN NAIROBI, KENYA.

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Abstract

Ectodermal dysplasia (ED) is defined as a rare hereditary disorder involving two or more of the ectodermal structures, which include the skin, hair, nails, teeth, and sweat glands. The two most common forms of the disease are hypohidrotic/anhidrotic ED and hidrotic ED.

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ABSTRACT.

Ectodermal dysplasia (ED) is defined as a rare hereditary disorder involving two or more of the ectodermal structures, which include the skin, hair, nails, teeth, and sweat glands. The two most common forms of the disease are hypohidrotic/anhidrotic ED (Christ-Siemens-Touraine syndrome) and hidrotic ED (Clouston's syndrome). The incidence of ED is 1 in 100,000 live births with the aetiology being attributed to a gene mutation.[1]

This case report presents the features, classification and management of a 2-year-old presenting with hypohidrotic ectodermal dysplasia in a private dental clinic in Nairobi, Kenya

INTRODUCTION.

Ectodermal dysplasia is a rare genetic disorder characterised by the presences of a congenital defect of 2 or more tissues derived from the embryonic ectoderm with the tissues affected being teeth, skin and its appendages.[2] There are currently more than 192 ED syndromes with varying modes of inheritance.

ED can occur as early as the 6th week of intrauterine life and during the first trimester. There are two major classifications of the disorder which is based on number and functionality of sweat glands. First type is anhidrotic/hypohidrotic ED (Christ-Siemens-Touraine syndrome); whereby sweat glands are significantly reduced or absent. The second type is the Hidrotic ED (Clouston's syndrome); whereby the sweat glands are normal.[3]

Hypohidrotic ED is more common and is usually inherited as an X-linked recessive trait hence exhibiting a male preponderance.[4] The earliest report of a patient with ectodermal dysplasia was published in 1848 by

Thurnam et al. and the term ectodermal dysplasia was coined in 1929 by Weech et al.[5]

CASE REPORT

A mother presented her 2-year-old son to the dental clinic. The presenting complaint was that her son had very few teeth for his age and they were extremely pointy and sharp. As a result, they were causing serious injury to his younger brother when biting him during play. The parent requested to have the few teeth to be blunted and to have the missing teeth replaced. In addition, the mother was distraught with concern over the child's aesthetics and poor feeding due to the lack of teeth. Family history revealed that the disorder was present in some of the patient's maternal cousins and some of his maternal uncles. General examination revealed that the patient was responsive, conscious and pleasant, however, patient was non-cooperative on the dental chair. Extra-orally the patient had frontal bossing, sunken cheeks, a saddle nose, thick protruded everted lips, large low set ears, wrinkled and hyperpigmented skin around the eyes. The hair on the scalp was sparse, short and fine (Figure 1 and 2). His nails were thin, brittle appearing with abnormal ridging on the toe nails (Figure 5 and 6). Intraorally there was dry mucosa indicative of xerostomia. Hypodontia was present with only tooth number 51 and 61 conically shaped crowns being visible clinically. The alveolar ridge was knife shaped indicative of congenitally missing teeth (Figure 3). An Orthopantomogram (Figure 4) showed typical conical teeth and congenital hypodontia. With no reported history of parental consanguinity and the disorder presentation being more pronounced in males from the patient's maternal side the authors labelled the ED as an X-linked recessive ED.[6]

Oral hygiene instructions were given and together with diet counselling. Genetic counselling was given and mother educated about the condition and the dental effects of xerostomia. Fluoride application was done using 1.23% Acidulated Phosphate Fluoride. The immediate management provided was to blunt the conically shaped teeth (51 and 61) to minimize the possibility of trauma to self or to others. Using a high-speed handpiece and a white-stone bur, the incisal tips were slightly rounded to reduce the sharpness. This was repeated after 6 weeks, to minimize the risk of sensitivity. The missing teeth will be replaced by a dental prosthesis which would be fabricated after further growth of the child and subsequently further eruption.

DISCUSSION

Hypohidrotic ectodermal dysplasia (HED) is usually transmitted as an X-linked recessive trait where females are carriers and is manifested in males. The unaffected female has 50% chance of transmitting this disorder to both her male and each female child. Female offspring have a 50% chance of inheriting the defective gene, thereby being a carrier. There is minimal expression in female carriers in the form of hypodontia and or conical teeth and reduced sweating.[6] There is possibility of spontaneous gene mutation where it may occur in family without any history of the syndrome. The prevalence in the population ranges between 1:10,000 and 1:100,000 male live births.[4]

Clinical features of ectodermal dysplasia include: hypohidrosis where there is decreased sweating than usual as the eccrine glands may be absent, sparse or rudimentary. This often affects the patient's heat tolerance. Onychodysplasia is common where there is congenital non hereditary nail disorder. Nail defects may differ by syndrome for example nails for those with Clouston's syndrome can be thick and discoloured with slow growth while those with hypohidrotic ectodermal dysplasia experience thin, brittle nails.[7]Hypotrichosis is seen where the hair on the scalp and eyebrows is sparse and fine in texture. Excessive drying and scaling of the skin is due to diminished number of sebaceous glands therefore reduced sebum production (asteatosis).[3] Typical facial profile of the patient includes frontal bossing, sunken cheeks and a saddle nose; midface hypoplasia is evident often resulting in thick, protruded and everted lips, the skin around the eyes is wrinkled and hyperpigmented, and large, low-set ears, retruded appearance of the midface and reduced vertical dimension of face.

Patients with ectodermal dysplasia exhibit characteristic intraoral features. There may be total absence of teeth (anodontia) or partial absence of teeth (hypodontia) which are conically or peg shaped. This results in decreased vertical dimension. The alveolar ridge is not fully developed and high palatal arch and cleft palate may occur. Intraoral accessory glands may be hypoplastic resulting in xerostomia and consequently

dry and cracked lips. Lexner et al. reports that affected males will present with severe oligodontia while female carriers are likely to have milder hypodontia. [8]

Other common features are seen due to aberrations in keratinocyte function such as ophthalmic diseases, impaired lacrimal gland function and glaucoma. Respiratory health related issues such as foul-smelling nasal discharge, respiratory infections, recurrent pneumonias and wheezing, recurrent sinus are largely reported. This is caused when mucous glands are absent in the upper respiratory tract, bronchi and esophagus. [9]

Although timely diagnosis of HED can be difficult due to absence of hair and teeth at birth, dental treatment should start as soon as possible so as to minimize chances of abnormalities in speech and detrimental oral habits. It should seek to restore function and esthetics. This is often done as early as 3 years where prosthesis can be fabricated and later on implants can be considered so as to improve quality of life.[10]

CONCLUSION

A multidisciplinary approach is important for the definitive management of patients affected by ectodermal dysplasia. Dental treatment is customized for every patient at different ages and often times requires long term care and may need complex procedures to ensure the patient has improved oral health related quality of life.

REFERENCES.

1. Saraswarthi G and Sai A. 2017. Ectodermal dysplasia Report of 2 cases and review of literature. International Journal of current research 9(2): 47073-47077.
2. Freire-Maia N. 1977. Ectodermal dysplasias revisited. Acta geneticae medicae et gemellologiae: twin research 26(2):121-131.
3. Pandey R, Khatri A. 2017. Dental Management of Ectodermal Dysplasia: A Report of Two Cases. Indian J Dent Adv 9(3):191-196.
4. Deshmukh S, Prashanth S. 2012. Ectodermal dysplasia: a genetic review. International Journal of Clinical Pediatric Dentistry 5(3):197.
5. Weech AA. 1929. Hereditary ectodermal dysplasia (congenital ectodermal defect): a report of two cases. American Journal of Diseases of Children 37(4):766-790.
6. Pinheiro M, Freire-Maia N. 1994. Ectodermal dysplasias: A clinical classification and a causal review. American journal of medical genetics 53(2):153-162.
7. National Foundation of Ectodermal Dysplasia. Accessed via: Symptoms - NFED.
8. Lexner MO, Bardow A, Hertz JM, Nielsen LA, Kreiborg S. 2007. Anomalies of tooth formation in hypohidrotic ectodermal dysplasia. International Journal of Paediatric Dentistry 17(1):10-18.
9. Fete M, Hermann J, Behrens J, Huttner KM. 2014. X-linked hypohidrotic ectodermal dysplasia (XL-HED): Clinical and diagnostic insights from an international patient registry. American Journal of Medical Genetics Part A 164(10):2437-2442.
10. National Foundation of Ectodermal Dysplasia. Accessed via: Dental Treatment Options - NFED.

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