Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/mjafi

Case Report

Hypohidrotic Ectodermal Dysplasia

Capt Gaurav Pratap Singh ^{a,*}, Lt Col Vivek Saxena ^b^a Dental Officer, 327 Fd Hospital, C/O 99 APO, India^b Classified Specialist (Oral & Maxillofacial Surgery), Command Military Dental Centre (Central Command), Lucknow, UP, India

ARTICLE INFO

Article history:

Received 24 December 2013

Accepted 25 May 2014

Available online 5 August 2014

Keywords:

Ectodermal dysplasia

X-linked recessive

Oligodontia

Hypotrichosis

Anhidrosis

Introduction

Ectodermal dysplasias are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. These tissues include the skin, hair, nails, eccrine glands and teeth.¹ Hypohidrotic Ectodermal Dysplasia is the most common type and is usually inherited as an X-linked recessive trait. It is characterised by the triad of signs which comprises 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 incidence of ectodermal dysplasia in males is estimated at 1 in 100,000 births.²

The aim of this case report is to examine a rare and unusual case of familial hypohidrotic ectodermal dysplasia, study the signs and symptoms of this condition and review the diagnostic and treatment modalities to be followed.

Case report

A 5 year old child patient reported to dental OPD with a chief complaint of missing teeth in both upper and lower arch. The patient gave a history of missing teeth since birth and a history of intolerance to heat. No other associated medical or systemic conditions were present. On extra oral examination patient gave a characteristic appearance of sparse scalp hair (fine and dry), frontal bossing, sunken cheeks, saddle nose, thick everted lips and large low set ears (Fig. 1). On intra oral examination, patient presented with oligodontia with presence of deciduous second molars and deciduous maxillary central incisors (caries) (Fig. 1). He also presented with a history of intolerance to heat suggesting sparse or rudimentary eccrine sweat glands. Mother and father were both clinically normal with no presenting symptoms. The patient had two male siblings aged 10 yrs and 3 yrs respectively. He was advised an Orthopantomogram (OPG) and recalled after two days along with his siblings.

On the next appointment, the younger sibling also exhibited similar extra oral and intra oral features (Fig. 2). The older sibling was however clinically normal. On further enquiry, no other family members exhibited similar symptoms. There was no history of consanguineous marriage of parents. OPG revealed absence of permanent tooth buds with the exception of permanent first molars (Fig. 3). Based on the clinical and radiographic findings, a diagnosis of Hypohidrotic Ectodermal Dysplasia was made.

Discussion

The Ectodermal Dysplasias are a group of inherited disorders that share in common developmental defects involving at

* Corresponding author. Tel.: +91 9475230995.

E-mail address: gaurav3000ad@gmail.com (G.P. Singh).<http://dx.doi.org/10.1016/j.mjafi.2014.05.006>

0377-1237/© 2014, Armed Forces Medical Services (AFMS). All rights reserved.

Fig. 1 – Extra oral image showing characteristic facies with sparse hair, frontal bossing, saddle nose, low set ears, everted lips and scaly skin. Intra oral view showing presence of oligodontia, deformed teeth and presence of caries. Fingers and toes show no abnormality. No evidence of ridging or deformed nails.

least two of the major structures classically derived from the embryogenic ectoderms including hair, teeth, nails, sweat glands. It was first described by Thumam in 1848 and was coined by Weech in 1929.³ Etiology is genetic.⁴ Usually Ectodermal Dysplasia is divided into two types based on the number and function of sweat glands as Hypohidrotic (anhidrotic) Ectodermal Dysplasia (Christ–Siemens–Tourine syndrome) in which sweat glands are absent or significantly decreased and Hidrotic Ectodermal Dysplasia (Clouston syndrome) in which the patients have normal facies, normal sweating and no specific defect is seen apart from nail dystrophy, hair defects and palmoplantar dyskeratosis.⁵

The differential diagnosis of hypohidrotic ectodermal dysplasia includes Rothmund–Thomson syndrome which exhibits features of sun-sensitive rash with prominent poikiloderma and telangiectasias, saddle nose, sparse hair growth, juvenile cataracts and hypodontia. However none of

the numerous cases of ectodermal dysplasias of the anhidrotic or hidrotic type reported in literature exhibited cataracts in infancy or in later life. Other conditions to be considered while diagnosing hypohidrotic ectodermal dysplasia include Alopecia Areata, Aplasia Cutis Congenita, Focal Dermal Hypoplasia syndrome, Incontinentia Pigmenti, Naegeli–Franceschetti–Jadassohn syndrome, Pachyonychia Congenita and Werner's syndrome.⁶

The extra oral features observed in our case were frontal bossing with the forehead appearing square in shape, prominent supra orbital ridge, depressed nasal bridge (saddle nose), everted lips, midfacial hypoplasia, low set ears and scaling of skin. The most characteristic intra oral findings which were observed included reduced number (oligodontia) and abnormal shape of teeth. Hypoplastic salivary glands results in xerostomia which is a cause for increased caries incidence in these patients. Other features include brittle nails with

Fig. 2 – 3 year old sibling also displaying similar extra oral features and presence of oligodontia.

abnormal ridging and missing fingers or toes. Occasionally cleft lip and/or palate is also seen.¹

Prenatal diagnosis of ectodermal dysplasia has occasionally been reported, which has been diagnosed by foetal skin

Fig. 3 – OPG revealing absence of permanent tooth buds with the exception of permanent first molars.

Fig. 4 – X-linked recessive mode of inheritance.

biopsy, obtained through foetoscopy by 20 weeks of gestation.⁷ The characteristic facies is pathognomonic but may not be recognized in infancy. In partial forms, the pointed conical teeth provide the most valuable indication and should suggest the need for sweat test and a skin biopsy.⁸

Genetic studies of more than 300 cases have revealed X-linked mode of inheritance (Fig. 4) with its gene locus being Xq11-21.1. The gene is carried by the female but manifested in the male. The complete syndrome does not occur in females but females may show dental defects, sparse hair, reduced sweating and dermatoglyphic abnormalities.⁸ The same pattern was observed in our case where the mother was clinically normal but two out of three male children exhibited features of ectodermal dysplasia. Autosomal recessive and autosomal dominant forms have been reported but are rare.¹

The management of children and adults with HED is a challenge because of their heat intolerance (especially during febrile illness or physical activities and in warm climate) and because of their susceptibility to pulmonary infections. During hot weather, affected individuals must have access to an adequate supply of water and a cool environment, which may mean “cooling vests,” air conditioning, a wet T-shirt, and/or a spray bottle of water. However, external cooling is less effective in these patients because their heat transfer from the core to the skin is also reduced, presumably due to poor capillary dilatation.⁹

The most frequent prosthetic treatment for the dental management of ectodermal dysplasia is removable prosthodontics due to ongoing growth and development and insufficient alveolar bone support.¹⁰ Since alveolar bone development is dependent on the presence of teeth, children with ectodermal dysplasia have little or no bone ridge upon which to construct dentures; therefore, restoring function and appearance is more challenging than usual.⁵ Dental implants combined with implant-supported dentures for adolescents

over 12 years of age are recommended as a treatment choice in literature. However if bone atrophy progresses in these already alveolar-deficient patients, implant placement may not be possible without bone grafting.¹¹

In patients diagnosed with Ectodermal Dysplasia, emphasis should be on recognition and early diagnosis of the condition which should be followed by a thorough family history and clinical examination of family members for similar signs and symptoms. There is no treatment for the condition,¹ however affected individuals with dental defects could be provided early dental evaluation and intervention. Prosthetic treatment should aim at early restoration of function and esthetics. Regular follow up is a must, not only to make provision for the growing dental arch and other dental problems but to holistically evaluate this condition and referral for medical opinion as and when required.

Conflicts of interest

All authors have none to declare.

REFERENCES

1. Rajendran R. *Shafer's Textbook of Oral Pathology*. 5th ed. Philadelphia: Saunders; 1983:797–798.
2. Stevenson AC, Kerr CB. On the distribution of the frequencies of mutation to genes determining harmful traits in man. *Mutat Res*. 1967;4:339–352.
3. Neopane A, Singh SK, Manandhar B. Hypohidrotic ectodermal dysplasia. *Kathmandu Univ Med J*. 2006;16:520–521.
4. Mortier K, Wackens G. Ectodermal dysplasia anhidrotic. *Orphanet Encycl*. 2004:1–6.
5. Vieira KA, Teixeira MS, Guirado CG, Gavião MB. Prosthodontic treatment of hypohidrotic ectodermal dysplasia with complete anodontia: case report. *Quintessence Int*. 2007;38:75–80.
6. Thannhauser SJ. Differential diagnosis of ectodermal dysplasia and of Rothmund's syndrome. *J Am Med Assoc*. 1946;130(4):238.
7. Bergendal B, Koch G, Kurol J, Wänndahl G. *Consensus Conference on Ectodermal Dysplasia with Special Reference to Dental Treatment*. Jönköping, Sweden: The Institute for Postgraduate Dental Education; 1998.
8. Anoop TM, Simi S, Mini PN, et al. Hypohidrotic ectodermal dysplasia. *J Assoc Physicians India*. 2008;56:268–270.
9. Brengelmann GL, Freund PR, Rowell LB, Olerud JE, Kraning KK. Absence of active cutaneous vasodilation associated with congenital absence of sweat glands in humans. *Am J Physiol*. 1981;240:571–575.
10. Pigno MA, Blackman RB, Cronin RJ, Cavazos E. Prosthodontic management of ectodermal dysplasia: a review of the literature. *J Prosthet Dent*. 1996;76:541–545.
11. Imirzalioglu P, Uckan S, Haydar SG. Surgical and prosthodontic treatment alternatives for children and adolescents with ectodermal dysplasia: a clinical report. *J Prosthet Dent*. 2002;88:569–572.