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Case Report

Hypohydrotic Ectodermal Dysplasia: A Case Report and Literature Review

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Abstract: Ectodermal dysplasia is a hereditary disorder that occurs as a consequence of disturbances in the ectoderm of the developing embryo. In the triad of alopecia or hypotrichosis and palmoplantar hyperkeratosis, nail dystrophy is usually accompanied by a lack of sweat glands and a partial or complete absence of primary and/ or permanent dentition. Here, we are presenting a rare case of Ectodermal dysplasia in a 7-year female with classic features of this condition.

Keywords: Ectodermal dysplasia, embryo, palmoplantar hyperkeratosis.

INTRODUCTION

Ectodermal dysplasia is a rare hereditary congenital disease that affects several ectodermal structures. This group of rare inherited disorders is due to developmental disturbances in the embryonal stage. The outer layer of cells in a developing embryo consists of an ectoderm. Structures formed from ectoderm include the teeth, epidermis and appendageal structures, the nervous system, and the organs of special senses [1]. Mutations in the EDA, EDAR, and EDARADD genes cause HED. EDA is the only gene known to be associated with X-linked HED (XLHED). Ninety-five percent of individuals with HED have the X-linked form. The genes EDAR and EDARADD are known to be associated with both autosomal dominant and autosomal recessive forms of HED. Mutations in the EDA, EDAR or EDARADD gene results in defective ectodysplasin A formation thereby preventing normal interactions between the ectoderm and the mesoderm and hence impairing the normal development of hair, sweat glands and teeth. The improper formation of these ectodermal structures leads to the characteristic features of hypohidrotic ectodermal dysplasia.

Children with Ectodermal Dysplasia (ED) may have various manifestations of the disease which may or may not involve teeth, skin, hair, nails, sweat glands, sebaceous and salivary glands. Ectodermal dysplasia may be defined as conditions with at least one of the following features Trichodysplasia, dental defects, onychodysplasia, or dishidrosis as well as at least one sign showing involvement of other ectodermal structures [2]. The most common syndromes within this group are hypohydrotic (anhidrotic) ED and hidrotic ED.

Hypohidrotic Ectodermal Dysplasia (HED) is manifested as a triad of defects that includes hypohidrosis, hypotrichosis, hypodontia. Patients with HED usually have skin that is soft, thin, dry, either with complete or partial absence of sweat glands. As a result, they cannot perspire normally and may have heat intolerance and hyperplasia. Hair follicles and sebaceous glands are often defective or absent and the hair of the scalp and eyebrows tends to be fine, scanty, and blond. Oral manifestations of HED include the partial or complete absence of teeth. This can affect both primary and permanent dentition. The teeth which are present may be conical in shape with large pulps, sometimes hypoplastic and delayed in eruption [3]. Oligodontia or hypodontia leads to atrophy of the alveolar bone. The permanent dentition may also present with impactions and transpositions [4]. In some cases, mucous glands are absent in the upper respiratory tract and the bronchi, esophagus, and duodenum. Other common signs are short stature, eye abnormalities, decreased tearing, and photophobia [5].

CASE REPORT

A seven-year-old female child reported to the Department of Oral Medicine and Radiology at Kamineni Institute of Dental Sciences, Narketpally with the chief complaint of multiple missing teeth in relation to upper and lower arch. The patient presented no history of exfoliation or extraction of teeth but gave a history of delayed eruption of teeth. The child was unable to tolerate hot climates and the parents revealed that the child used to wear wet clothes in summer to combat the heat. On examination, her hair was blond, fine, and sparsely distributed skin smooth and dry with wrinkles. The supraorbital ridges were prominent with sparse eyebrows. Nail examination revealed no abnormality. The lower facial height decreased due to over closed profile, making lips more prominent and protuberant (Fig 1). On intraoral examination, multiple missing teeth in the maxillary and mandibular arch, Maxillary central and lateral incisors appeared to be cone-shaped teeth, the color of alveolar mucosa and other oral mucosa was normal (Fig 2).

Fig 1: Extraoral photograph

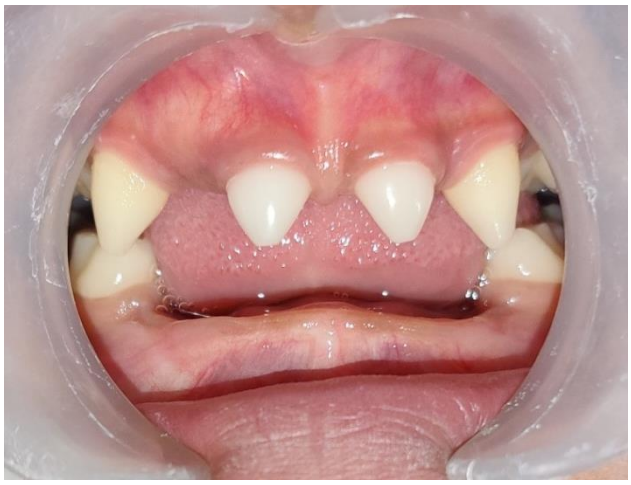

Fig 2: Intraoral photograph

Orthopantomogram (OPG) (Fig 3) was taken which revealed multiple missing teeth, aplasia of the alveolar processes of the maxilla and the mandible.

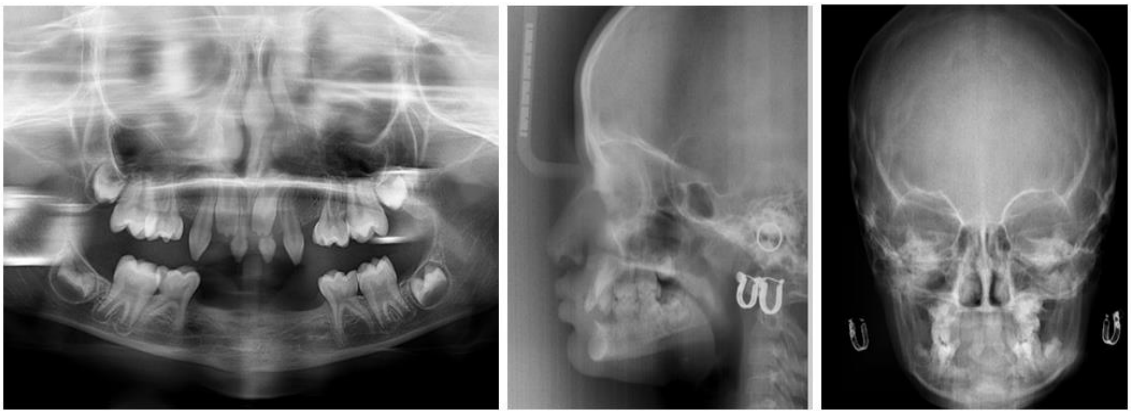

Fig 3: Radiographs showing multiple missing teeth & Alveolar ridges rather atrophic

DISCUSSION

The EDs are a heterogeneous group of hereditary disorders that occur approximately one in every 100,000 births which are caused by primary developmental defects of two or more embryonic ectoderm-derived tissues [6]. ED is divided into two broad categories, i.e., hypohidrotic form (Christ Touraine Syndrome) which is X-linked and is characterized by the classical triad of hypodontia, hypotrichosis, and hypohydrosis and the other category, i.e., hydrotic form (Clouston syndrome), which also affects the teeth, hair, and nails sparing the sweat glands. Extraorally fine, sparse, lusterless fair hair is seen over the scalp along with extensive scaling of the skin, and unexplained pyrexia and heat intolerance most commonly occurs due to anhidrosis. Normal intelligence is observed [7]. The other extraoral features are frontal bossing, sunken cheeks, depressed nasal bridge, thick everted protuberant lips, wrinkled hyperpigmented periorbital skin, and a large low set of ears. Hypodontia or anodontia of deciduous and permanent dentition associated with conical-shaped teeth is the most common oral symptom. Optimal treatment for children with hereditary ectodermal dysplasia requires a multidisciplinary approach. The patient reported here had involvement of hair, sweat glands, frontal bossing and she also had conical shaped teeth. These clinical features were supportive in diagnosing hydrotic ED. Intraorally missing permanent teeth are most commonly present, the maxillary central incisors and canines present with a conical crown form. In rare instances, one or both jaws may be edentulous, and the alveolar processes may not develop due to the absence of teeth.

CONCLUSION

EDs are rare genetic disorders that have many overlapping features, and it is difficult to classify them. The clinical manifestations of ED cause significant social problems in affected individuals. It disturbs both the oral functions and normal body functions of the patients. The key to success for the management of ED is the quick diagnosis and prosthetic rehabilitation by the multidisciplinary approach.

REFERENCES

1. Shashibhushan, K. K., Viswanathan, R., Sathyajith Naik, N., & Subba Reddy, V. V. (2011). Hypohidrotic ectodermal dysplasia with total anodontia: A case report.
2. Ramos, V., Giebink, D. L., Fisher, J. G., & Christensen, L. C. (1995). Complete dentures for a child with hypohidrotic ectodermal dysplasia: a clinical report. *The Journal of prosthetic dentistry*, *74*(4), 329-331.
3. Crawford, P. J., Aldred, M. J., & Clarke, A. (1991). Clinical and radiographic dental findings in X linked hypohidrotic ectodermal dysplasia. *Journal of medical genetics*, *28*(3), 181-185.
4. Borg, P., & Midtgaard, K. (1977). Ectodermal dysplasia: report of four cases. *ASDC journal of dentistry for children*, *44*(4), 314-319.
5. Chee, S. Y., Wanga, C. H., Lina, W. D., & Tsaia, F. J. (2014). Ectodermal dysplasia (ED) syndrome. *BioMedicine*, *4*(4), 1-2.
6. Clarke, A. (1987). Hypohidrotic ectodermal dysplasia. *Journal of medical genetics*, *24*(11), 659-663.
7. Crawford, P. J., Aldred, M. J., Clarke, A., & Tso, M. S. (1989). Rapp-Hodgkin syndrome: An ectodermal dysplasia involving the teeth, hair, nails, and palate: Report of a case and review of the literature. *Oral surgery, oral medicine, oral pathology*, *67*(1), 50-62.