Hidrotic Ectodermal Dysplasia: Case Report and Review

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ABSTRACT

Ectodermal dysplasia is a group of disorders defined by the abnormal development of two or more structures derived from the ectodermal layer. Patients with ectodermal dysplasia are characterized by hypoplasia or aplasia of structures such as skin, hair, nails, teeth, nerve cells, sweat glands, parts of the eye and ear and other organs. It is divided into two major groups: Hypohidrotic and Hidrotic. The gene that causes hidrotic ectodermal dysplasia (Clouston's syndrome) has been identified to be the GJB6, which encodes for connexin-30. GBJ-6 has been mapped to the pericentromeric region of chromosome 13q. Inheritance is autosomal dominant, but there may be considerable variation in expression. Severity is more pronounced in males than in females and thus females show only minor defects. Common features include spare hair on scalp and other part parts, dry skin, intolerance to heat, oligodontia or complete anodontia, prominence of frontal bone and supra orbital ridges, spoon shaped or concave nails. Carriers present with very less symptoms when compared to a patient which is actually affected. So we present a case of a hidrotic ectodermal dysplasia affecting a female patient so that the features and review can be highlighted.

Key words: *Ectodermal dysplasia, Hidrotic, Oligodontia, midface hypoplasia*

INTRODUCTION

Ectodermal dysplasia is a group of disorders defined by the abnormal development of two or more structures derived from the ectodermal layer. The ectoderm, one of three germ layers present in the developing embryo, gives rise to the central nervous system, peripheral nervous system, sweat glands, hair, nails, and enamel of the teeth.[1] The earliest recorded cases of ectodermal dysplasia were described in 1792. Since then, more than 200 different pathologic clinical conditions have been recognized and defined as ectodermal dysplasia. These disorders are considered relatively rare, 1 in 10,000 –1 in 100,000 births.[2] Patients with ectodermal dysplasia are characterized by hypoplasia or aplasia of structures such as skin, hair, nails, teeth, nerve cells, sweat glands, parts of the eye and ear and other organs.[3]

According to the state of sweat glands involvement, two major groups are distinguished: 1) Hypohidrotic or anhidrotic (Christ-Siemens-Touriane syndrome) in which sweat glands are either absent or significantly reduced in number; (2) Hidrotic (Cloustone syndrome) in which sweat glands are normal. Dentition and hair are involved similarly in both types but hereditary patterns of nails and sweat glands involvement are different. Hypohidrotic ectodermal dysplasia as the most common type seems to show an X-linked inheritance pattern with the gene mapping to Xq12-q13.[3] The disease is characterized by deformity of at least two or more of these tissues, which primarily involves skin, hair, nails, eccrine glands, and teeth, which makes it difficult for the patient not only to masticate food, but it also causes a psychological impact due to partial edentulism.[4] Here, we are presenting a rare case of Hidrotic Ectodermal Dysplasia with typical features in a 35 year old female.

CASE REPORT

A female patient aged 35 years came to the Department of Oral Medicine & Radiology with a chief complaint of missing upper and lower front teeth since childhood (*Fig.5*). Patient gave history of exfoliation of milk teeth at the age of 7 years and since then there is spacing in the upper and lower front teeth. Patient's family history revealed that her grandmother from maternal side also had a similar complaint. Extra- oral examination of patient revealed dry skin and spare hair on the arms and the legs. Further examination revealed that the hair of the scalp and eyebrows and the shape of the nails were normal. Examination of forehead revealed prominent forntal bone and the supraorbital ridges (*Fig 1,7*). The lower lip of the patient was protruded and dry. Intra – oral examination of patient revealed missing teeth (15,17,25,27,31,32,37,41,42,47) (*Fig 3,4*). The crowns of the maxillary lateral incisors were conical in shape and the occlusal anatomy of the maxillary first molars was similar to maxillary first premolars. Also patient was having a high palate and deep bite.

Patient was then referred for radiographic investigation. OPG (*Fig 6*) investigation revealed no impacted teeth, the roots of the teeth present were short and conical in shape, oligodontia was seen with absence of tooth germ of the missing teeth. Lateral

Cephalogram (*Fig 7*) revealed prominent frontal and occipital bone, reduced mid – facial height and hypoplastic maxilla.

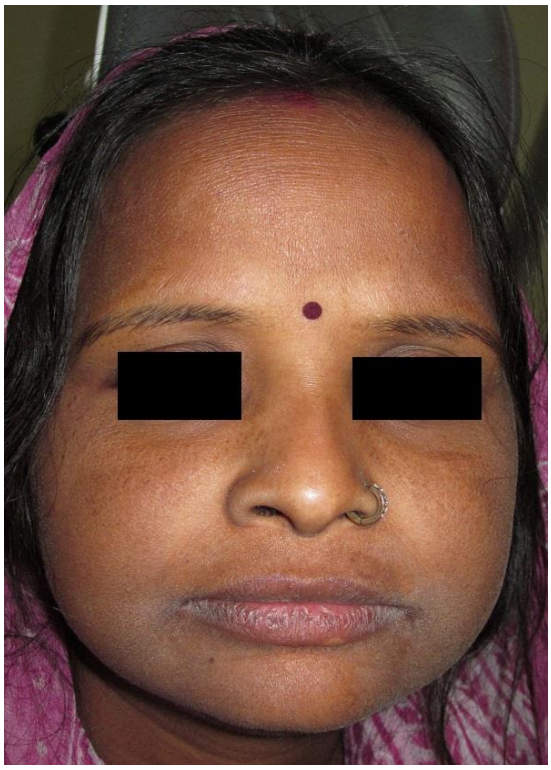

Fig. 1: Facial Profile

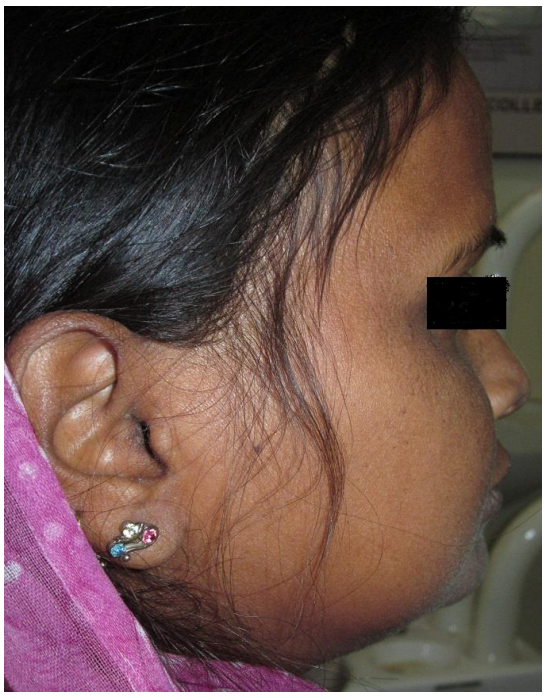

Fig. 2: Side Profile

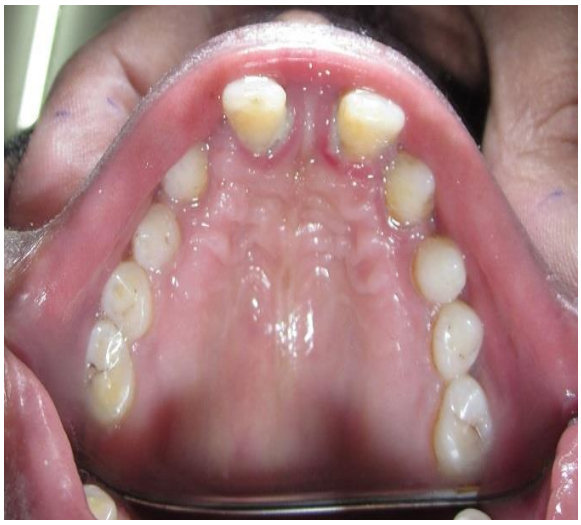

Fig. 3: Maxillary Arch

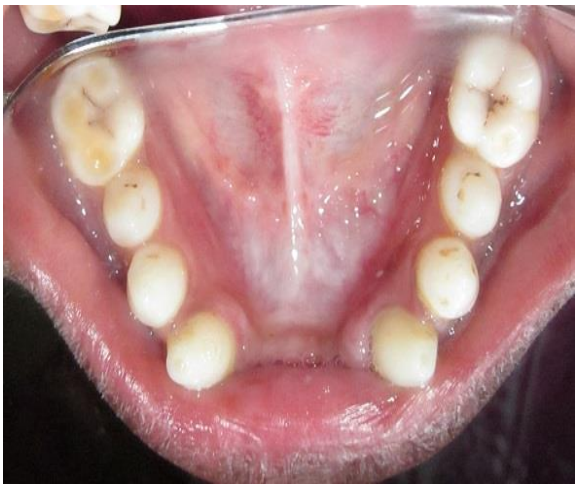

Fig. 4: Mandibular Arch

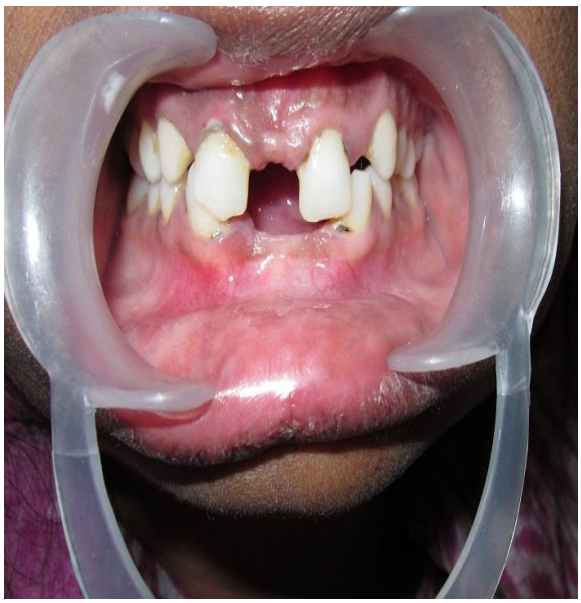

Fig. 5: Anterior Missing Teeth

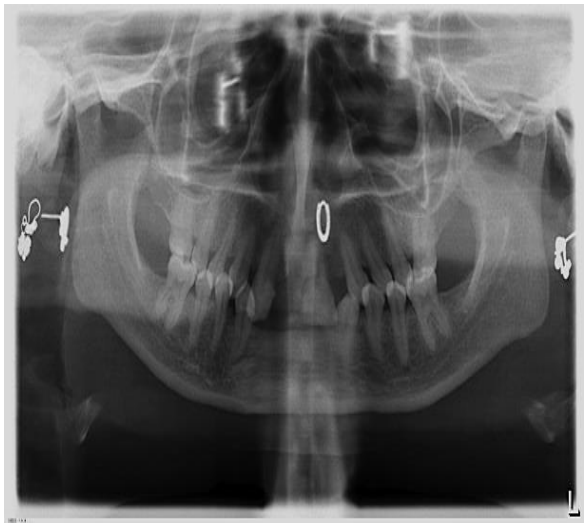

Fig. 6: OPG of Patient

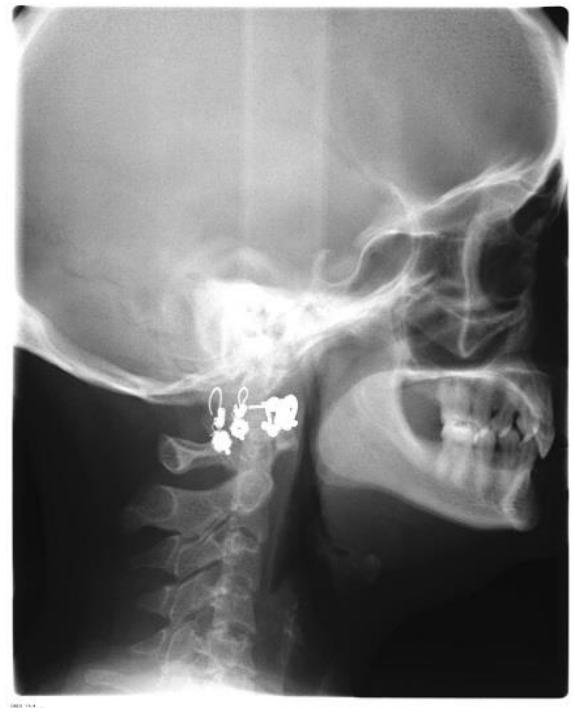

Fig. 7: Lateral Cephalogram of Patient

DISCUSSION & REVIEW

Ectodermal Dysplasia is a disorder that might occur during the first trimester of pregnancy. If it is severe, it appears before the sixth week of embryonic life and consequently the dentition will be affected. After eighth week other ectodermal structure may be affected.[5] It is typically inherited as a cross-linked recessive trait so that frequency and severity of this condition is more pronounced in males than in females and thus females show only minor defects.[6,7,10]

The gene that causes hidrotic ectodermal dysplasia (Clouston's syndrome) has been identified to be the GJB6, which encodes for connexin-30. GBJ-6 has been mapped to the pericentromeric region of chromosome 13q. Mutations of the gene PVRL1, encoding a cell-to-cell adhesion molecule/herpes virus receptor, have been reported in those with cleft lip/palate ectodermal dysplasia.[7]

Dental Abnormalities

The dental finding in ectodermal dysplasia may range from hypodontia to anodontia of the primary or permanent teeth. However congenital absence of primary teeth is relatively rare. Frequently, the teeth that are present have conical crowns.[8] In the present case there are expressions of these features which includes hypodontia in the permanent dentition and conical shape of maxillary lateral incisors.

Craniofacial Development

The presence of teeth produces alveolar thickening bur has no influence on the growth and development of the jaws which is normal in these patients. Because the alveolar process does not develop in the absence of teeth the vertical dimension is reduced in these subjects resulting in protuberance of lips. The palatal arch is frequently high with or without presence of cleft palate.[7] Further there have also been reports of presence of prominent supraorbital ridges and frontal eminence.[10] There are some example of patients having a hypoplastic maxilla which is relatively less common.[9] In the presenting case the apparent craniofacial features seen were underdeveloped alveolar process resulting in reduced vertical dimension and protruding lower lip, high arch palate and prominent supraorbial ridges and frontal eminence.

Skin

Most of the affected subjects have normal skin, but some have dry skin but none of the patients with dry skin gave a history of heat intolerance or problem owing to lack of sweating.[9] In the present case patient reportedly gave history of dry skin but did not reveal any history of heat intolerance.

Hair

The hair has a normal distribution, but may be fine, slow growing and sparse. Total alopecia has not been described. There is considerable variation in degree to which hair is affected, and some gene carriers have apparently normal hair or silky, soft hair.[11] Other body hair is not usually affected, but there are cases reported with sparse eye lashes and eyebrows.[12] In the present case no defect was seen with the hair of the patient neither with that of the scalp nor other body hair.

Nails

The nails are described as small and dysplastic and may be spoon shaped or concave. Nails are often

slow growing and the toe nails are usually more severely affected. As in case of teeth and hair, nail dystrophy shows considerable variations in gene carriers: in some there is no evidence of nail dystrophy, whereas in others only the toe nails are affected and to a very variable extent.[9] In the present case no evident nail dystrophy was noted.

Inheritance

Inheritance is autosomal dominant, but there may be considerable variation in expression. Careful examination of relatives of affected subjects must be performed before advising on carrier status, as gene carriers may have minimal signs.[9]

Management

Patients with hidrotic form do not present as many immediate problems. Dental and hair problems are common in both forms. Early dental consultation should be obtained whenever there is hypodontia in order that the child may be fitted with prosthesis.[10] When indicated, appropriate care needs to be rendered throughout the child's growth cycle to maintain oral functions as well as to address the aesthetic needs of the patient.

CONCLUSION

Hidrotic Ectodermal Dysplasia is an autosomal dominant condition with various features which include defect in hair, skin, nails, dental anomalies, craniofacial development. Moreover carriers in such cases present with very less symptoms when compared to a patient which is actually affected by such condition. More such cases should be reported so that the features that present in carriers of ectodermal dysplasia can be highlighted.

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