HYPOHIDROTIC ECTODERMAL DYSPLASIA: A CASE REPORT

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Ectodermal dysplasia is a hereditary disorder that occurs as a consequence of disturbance in ectoderm of developing embryo. Hypohidrotic ectodermal dysplasia or anhidrotic ectodermal dysplasia is most common syndrome among large group of hereditary disorders.the triad of nail dystrophy,alopecia or hypotrichosis and palmoplantar hyperkeratosis is usually accompanied by lack of sweat glands and a partial or complete absence of primary or permanent dentition. A case study of 39 year old female with hypohidrotic ectodermal dysplasia with positive family history is presented.

KEYWORDS: Ectodermal Dysplasia, Hypodontia, Anodontia, Hypohydrosis.

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INTRODUCTION

The Ectodermal dysplasia (EDs) comprises a large heterogeneous group of inherited disorders that are defined by primary defects in the development of 2 or more tissues derived from embryonic ectoderm. The tissues primarily involved are the skin, hair, nails, eccrine glands and teeth. Although Thurnam published the first report of a patient with ectodermal dysplasia in 1848,[1] but the term was not coined until 1929 by Weech[2].The ectodermal dysplaisas are congenital, diffuse and non-progressive disorder.

It comprises of over 150 different syndromes of unknown pathogenesis [3,4]. Hypohidrotic ectodermal dysplasia, also called as Christ-Siemens Touraine syndrome or anhidrotic ectodermal dysplasia is the most common of all forms. Its frequency has been reported to be affecting 1-7 per 10,000 live births [5].Clinical features include sparse, fine blond hair with abnormal texture of the scalp, eyebrows and eyelashes, dry skin, nail defects, prominent forehead, depressed nasal bridge and protuberant dry and cracked lips.[6,7,8,9].Complete, as well as partial, absence of sweat glands can cause dry skin and eczema and there may be heat intolerance or hyperthermia in warm condition[7,9]Xerostomia due to hypoplastic accessory salivary glands may cause drying and cracking of the lips[11]. The objective of this study is to report a case of hereditary Hypohidrotic ectodermal dysplasia in 39 yr old female.

CASE REPORT

A Thirty nine year old female from Mansehra reported to us in at OPD of ISLAMIC INTERNATIONAL DENTAL HOSPITAL with chief complaint of absence of teeth and wanted their replacement. She was referred to department of Prosthodontics. Dental history revealed delayed eruption of primary teeth and total absence of permanent teeth. She mentioned that only maxillary six teeth erupted out of which three exfoliated and three are still present. She also gave history of heat intolerance and almost complete absence of sweating.

Clinical examination of patient showed very fine, sparse and brittle hairs on the scalp, skin appeared to be dry and wrinkled. Depressed nasal bridge and periorbital pigmentation was also noted. (Figure 1).Frontal bossing with complete absence of eye brows and eyelashes was seen (Figure 2).The lower facial height is decreased due

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over closed profile making lips more prominent and protuberant. Nails appeared to be normal but skin of hands was dry and wrinkled. On dental examination total anodontia of mandible and maxillary hypodontia was seen. In maxillary arch two central deciduous incisors and left deciduous molar was seen (Figure3, 4). Orthopentanogram showed absence of teeth in mandible and presence of only primary central incisors and left

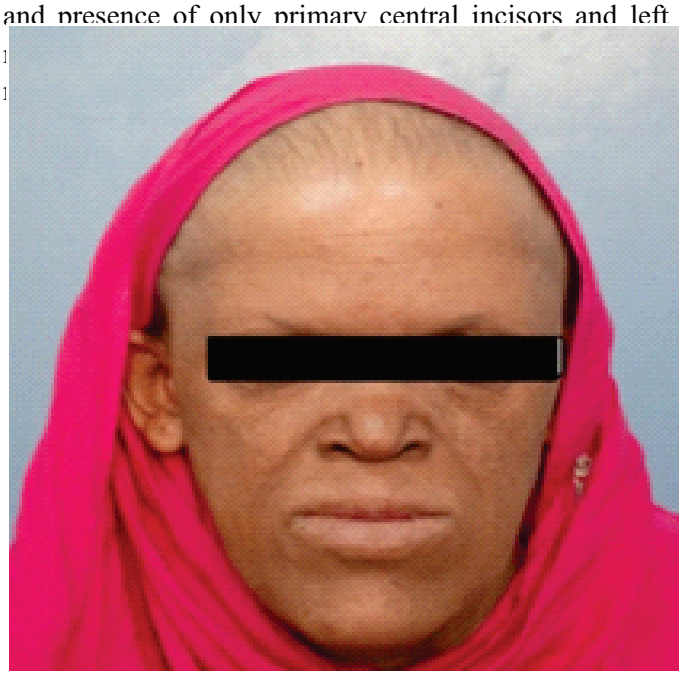

Fig.1 : 39 Year Old Lady with HED, Spares hair on scalp, depressed nasal bridge, protuberantlip, pigmentation around periobital region is shown

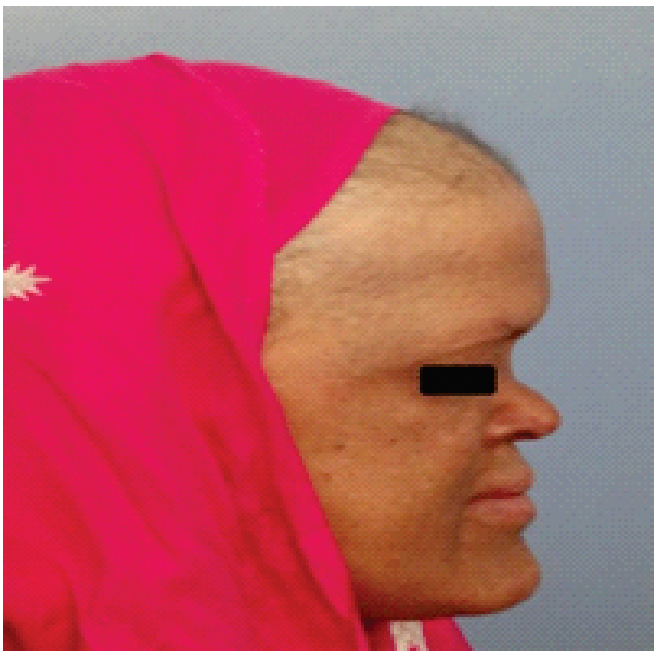

Fig.2 : Frontal bossing, decreased lower facial height

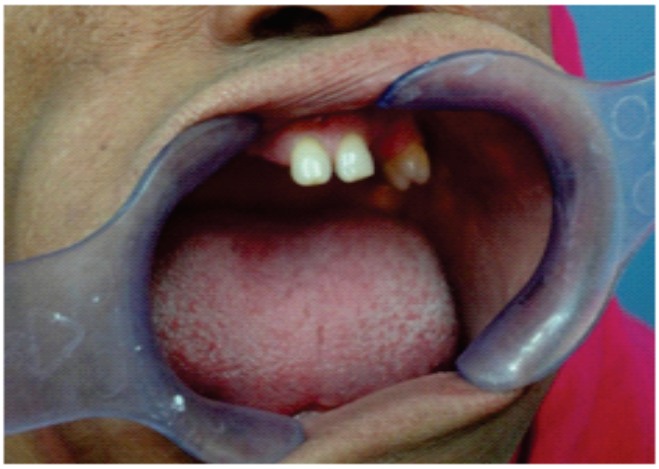

Fig.3 : Mandibular Anodontia

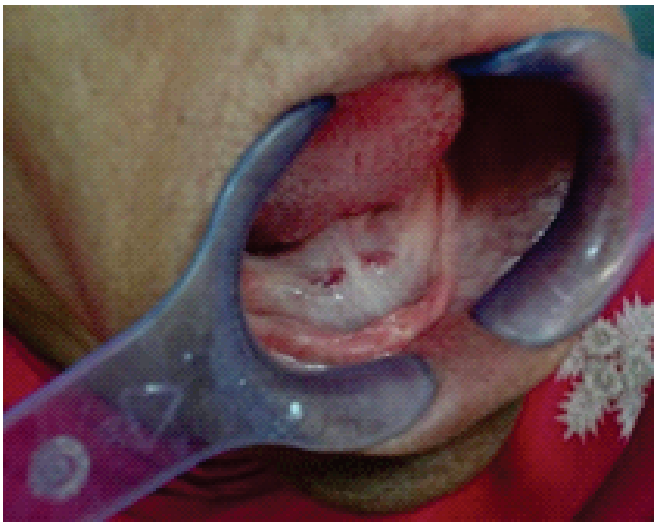

Fig.4 : Maxillary hypodontia with retained deciduous incisors and left molar

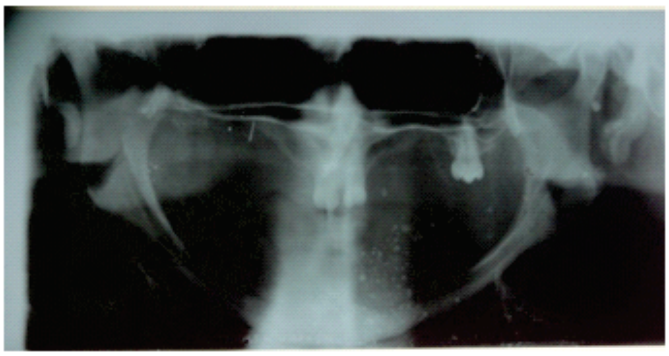

Fig.5 : OPG showing mandibular anodontia and maxillary hypodontia

The combined clinical and dental findings pointed towards the diagnosis of Hypohidrotic ectodermal dysplasia. She is youngest amongst her sister and brothers and she was only one affected amongst them. Her family

history also revealed that two of her niece and nephew are affected with this condition. Proposed treatment plan given to patient was mandibular complete denture and maxillary overdenture. But due to some of her personal issues she did not report to us for further treatment.

DISCUSSION

Usually the ectodermal dysplasiais divided into two types based on the number and function of sweat glands (Viera et al, 2007)[24] 1. Hypohidrotic ectodermal dysplasia / Christ-Siemens-Tourine Syndrome; where sweat glands are absent or markedly reduce in number. 2.Hidrotic Ectodermal Dysplasia /CloustonSyndrome; sweat glands are normal in number.

The dentition and hair are affected similarly in both types, but the hereditary patterns and nail and sweat gland manifestations tend to differ[12]**Hypohidrotic ectodermal** disorder is usually inherited as either autosomal dominant / recessive or X-linked recessive trait and the gene locus is X q13-q21.X-linked recessive Hypohidrotic ED (Christ-Siemens-Touraine syndrome), or anhidrotic ectodermal dysplasia and immunodeficiency (EDA-ID), is caused by mutations in gene EDAEDA, which encodes the ectodysplasin protein which in turn activates the NF-kappaB and essential modulator (NEMO), resulting in conical teeth, sparse hair, anhidrosis or Hypohidrotic, and recurrent bacterial infections[13].HED manifest as triad of hypohidrosis,hypotrichosisand hypodontia[14]. Characteristic features include frontal bossing reduction in amount of hair (hypotrichosis), absence of sweat glands (anhidrosis) resulting in temperature elevation, absence of sebaceous glands (asteatosis) resulting in dry skin, depressed nasal bridge, protuberant lips, prominent supra orbital ridges , sunken cheeks, wrinkled hyperpigmented skin around the eyes and large low set ears (Crawford et al ,1991).[15] .Oral traits of ectodermal dysplasia (ED) may be expressed as anodontia or hypodontia, with or without a cleft lip and palate. Anodontia also manifests itself by a lack of alveolar ridge development [16] as a result, the vertical dimension of the lower face is reduced, the vermilion border disappears, existing teeth are malformed, the oral mucosa becomes dry, and the lips become prominent. **Hidrotic ED (Clouston syndrome**), which is an autosomal dominant disorder, is caused by mutations in GJB6.The clinical features include nail dystrophy, hair defects and palmoplantardyskeratosis. The patients have normal facies, normal sweating and no specific defect is seen. The treatment for a patient with ectodermal dysplasia varies and generally depends on age of patient, dental agenesis, degree of malformation of teeth, the growth and development of the stomatognathic system of the patient and patient's motivation. For young patients suggested treatment include removable partial denture, complete denture ,over denture and flexible dentures implant therapy is not the treatment choice in children due to ongoing growth and development and insufficient alveolar bone support.[17].In adult patient all other treatment options can be pursued but the best are a fixed partial denture or and endosseous implant therapy. Lack of bone density, knife edge mandible ridge will require bone grafting for the placement of implants.[25,26]

Many case reports were presented regarding features of HED. Beckerman[10] reported the case of a female patient with anhidrotic ectodermal dysplasia and lacrimal gland anomaly. Shaw [8], Brodie and Sarnat[18]described cases of complete anodontia of both deciduous and permanent dentitions. .Mehmet Bani[19],reported two cases of ectodermal dysplasia with anodontia in young children..K Ramesh[20]reported 5 cases who suffered from Hypohidrotic ectodermal dysplasia. J Jananee [21] also presented acase on HED.In one case-controlled study of 68 persons with oligodontia, 57% had disturbances in hair, nails, and/or sweat production in addition to defective teeth, and were classified as having ED.[22]ArfanulBari[23] reported a case of HED involving sweat glands .Atopic eczema was seen on face and limbs. Other ectodermal structures like nails, hairs were unaffected. All teeth were present, just central incisors were malformed and widely spaced. This case appeared to be different from others. Medical care of patient depends on which ectodermal structure is involved. In above case report the temperature of body was elevated so it is advisable to have frequent consumption of cool liquids to maintain adequate hydration and thermocoagulation. For patient with dental defects, early dental evaluation and intervention is

advised. In above case report an overdenture in maxilla and in mandible complete denture or endosseous implants with ridge augumentation were advised.

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