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Case Report..

A Review and Report of 2 Cases of Ectodermal Dysplasia: A Clinical Portraiture

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ABSTRACT

Ectodermal dysplasia is a rare hereditary condition which mainly affects the structures of ectodermal origin including glands, tooth buds, hair follicles and nails. As the condition is characterised by hypodontia or anodontia which affects the teeth and dentist plays an important role in the diagnosis and rehabilitation of the patient. Patients feel difficulty in chewing, speaking, look different from others resulting in low self-esteem. Aim of the research is early diagnosis and reporting of the cases so as to refer the patients for rehabilitation to boost their self-confidence. Due to scarcity of research in this field, it becomes mandatory to put some light on the detailed clinical features of ectodermal dysplasia along with its hereditary correlation and clinical implications. We report two cases of hypohidrotic ectodermal dysplasia in a 12- and 25-years old boys with partial anodontia. A detailed clinical features along with hereditary relationship is explained along with signs and symptoms. In these cases, early diagnosis, prompt treatment and referral for rehabilitation are the keys to boost the self-esteem and restore the normal functions of the oral cavity. Hence the dentists should have a thorough knowledge of the diagnosis of such cases.

Keywords: Anodontia, Pseudorhagades, Midfacial Hypoplasia, Protuberant Lips.

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INTRODUCTION

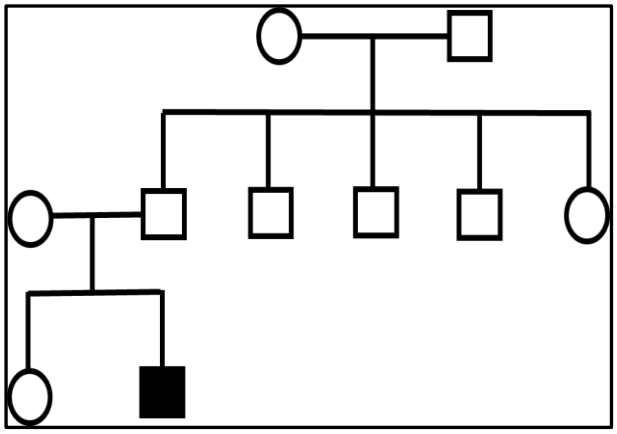
Ectodermal dysplasia is a rare hereditary condition which occurs as a consequence of disturbances information of ectodermal structures in the developing embryo.[1] It mainly affects glands, tooth buds, hair follicles and nail development. This syndrome was first described in literature by Thurnam, who reported two cases in 1848 followed by one case reported by Darwin in the 19th century. However, the term ectodermal dysplasia was not coined until 1929 by Weech.[2,3] Freire-Maia and Pinheiro proposed the first classification system of the ectodermal dysplasias in 1982,[4] with other updates in 1994 and 2001.[5,6] These are congenital, diffuse, nonprogressive and more than 192 distinct disorders have been described till date. Its estimated incidence is 1 to 2 in 10,000 births. Based on the number and functionality of sweat glands, two main types of ectodermal dysplasias have been characterised. These are X-linked recessive hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome), and hidrotic ectodermal dysplasia (Clouston syndrome).[1,7] Since the X-linked form is the most common, males are predominantly affected, although female carriers of an X chromosomal mutation can manifest partial symptoms in a mosaic pattern due to X-inactivation. Males and females are equally affected in the autosomal dominant and recessive forms of HED.[8]Most hypohidrotic ectodermal dysplasia cases are X-linked. XLHED is caused by mutations in the *EDA* gene, encoding the transmembrane protein ectodysplasin, a member of the tumor necrosis factor (TNF)-related ligand family involved in the early

epithelial-mesenchymal interaction that regulates ectodermal appendage formation.[8] main clinical features of this disorder include sparse, fine, with abnormal texture of the scalp, scanty eyebrows and eyelashes, dry skin, nail defects, frontal bossing, depressed nasal bridge and protuberant dry and cracked lips. Complete or partial absence of sweat glands which causes dry skin along with heat intolerance in warm conditions. Most common oral manifestations are anodontia or oligodontia with altered teeth morphology of both deciduous and permanent dentition. The roots of the teeth are usually short and conical. In case of complete anodontia, the alveolar ridge resorption is the most common finding with reduced vertical dimension leading to protuberant lips.[9]

CASE REPORT

CASE 1

A male patient of age 25 years of Indian origin visited the Department of Public Health Dentistry, SCB Dental College, Odisha, India, with the chief complaint of absence of teeth in his oral cavity since childhood with few teeth in maxillary arch and completely edentulous mandibular arch (fig 2b) along with dryness of mouth. There was no history of exfoliation or extraction of teeth but patient gave a history of delayed eruption of teeth. Patient was diagnosed with hypohidrotic ectodermal dysplasia after skin biopsy at SCB Medical College and Hospital. On clinical examination, intraorally the only teeth present were 17,16,11,21,26,27 (fig. 2a), incisors were cone-shaped, though all the other teeth also had altered morphology. The mandibular ridge was completely edentulous and appeared flat due to alveolar ridge resorption. The overall vertical dimension of face was also reduced. Orthopantomogram was taken to look for any impacted teeth. On radiographic examination no impacted teeth were observed, though the roots of teeth present were found to be short and conical. Patient also gave a history of dry mouth and dry skin with absence of sweat in his skin. He was intolerable to withstand hot water and hot environment. On extra oral examination, dry and scaly skin with slightly elevation of temperature was observed. Hair examination revealed fine sparse, lusterless appearance and eyebrows and eyelashes are absent with prominent supra-orbital ridges (fig.1). Ear examination revealed large low set, spoked ears, narrow face, prominent nasal bridge, upturned nose with midface hypoplasia gives an older look as compared with those of his age with a normal intelligence (fig.1). Patient had typical facies which was characterized by saddle nose, thick everted lips as well as thin, linear wrinkles in the peri-oral region which is hyperpigmented. There was no history of birth complications during his delivery, and no other live family member presented similar condition. None of the family members was involved in a similar manner in previous generations. There was no history of consanguinous marriage of parents. The systemic examination including otorhinolaryngological examination was normal. The physical development including external genitalia and mental development was normal. The routine biochemical tests were within normal limits. Diagnosis was confirmed with appropriate questionnaire, the detailed case history, intraoral and extraoral clinical examination revealing hypohidrosis, hypotrichosis, and hypodontia, radiographic examination and biopsy as these are the important tools to diagnose the Hypohidrotic ectodermal dysplasia.

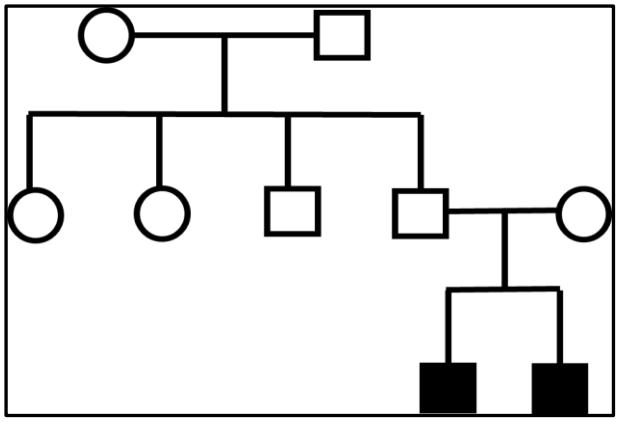


PEDIGREE FOR CASE 1

CASE 2

12 year old male patient reported to our institution with a chief complaint of missing teeth in upper and lower jaws since birth. On extra-oral examination the child demonstrated an array of peculiar features such as a receding hairline along with sparse light colored hair and scanty eyebrows (fig. 3 a, b, c). In general, the patient had dry and parched skin which gave a scaly appearance. The periorbital skin demonstrated a dry wrinkled appearance. Typical facial features such as a depressed nasal bridge, prominent supra-orbital ridges along with frontal bossing were present. His lips were thick everted protuberant with peri-oral region exhibiting increased pigmentation as well as thin, linear wrinkles (fig.3 a, b, c). His parent gave history of frequent bouts of fever and difficulty in tolerating high temperatures especially during summers with little tendency for sweating. His elder brother also suffered from similar condition as elicited by his family history. Ear examination revealed large low set ears with midface hypoplasia imparting a relatively older look for his age with a normal intelligence. On intra-oral examination dry mucous membrane as well as multiple missing teeth were noted with only 11, 21, 43 being present. However, the teeth present demonstrated an altered morphology being cone-shaped (fig.4a, 4b). Careful inspection showed thin relatively flat alveolar ridges, reduced vertical bone height, and loss of sulcus depth in the posterior regions of maxillary and mandibular jaws. The overall vertical dimension of face was also reduced.

Since hypohidrosis, hypotrichosis, and hypodontia were very evident on physical examination along with a positive family history, the patient was diagnosed with Hypohidrotic Ectodermal Dysplasia (HED) with partial anodontia.



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CASE 1

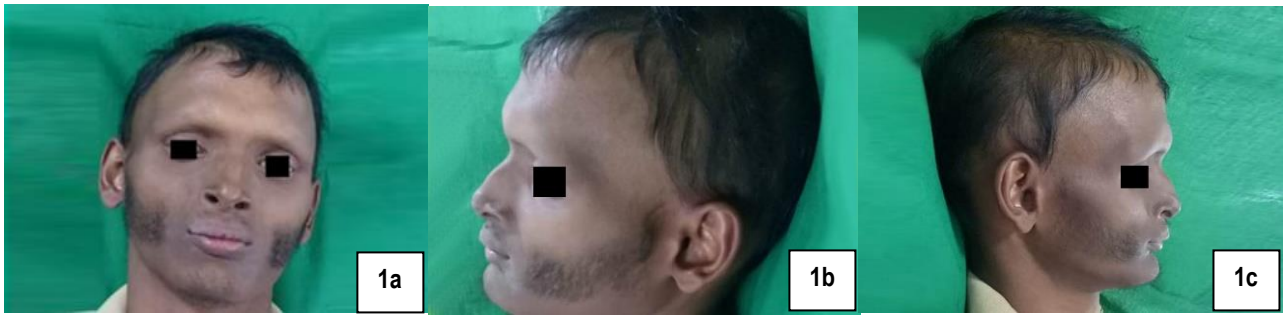

Figure 1 (1a,1b,1c): Extraoral profile views of case 1

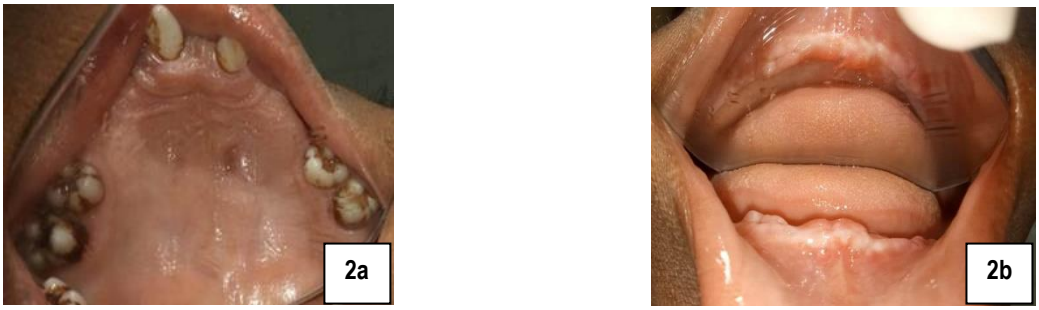

Figure 2 (2a, 2b): Intraoral photographs of case 1

CASE 2

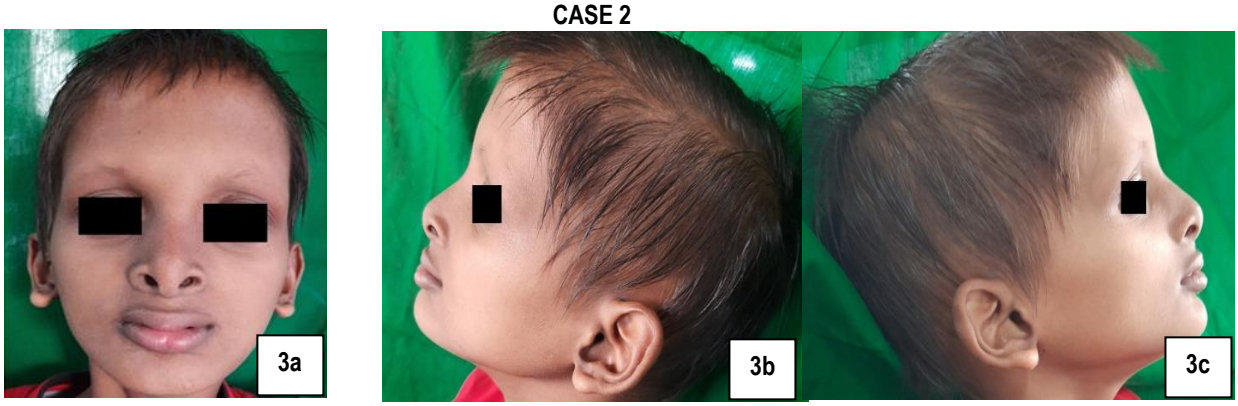

Figure 3 (3a, 3b, 3c)- Extraoral profile views of case 2

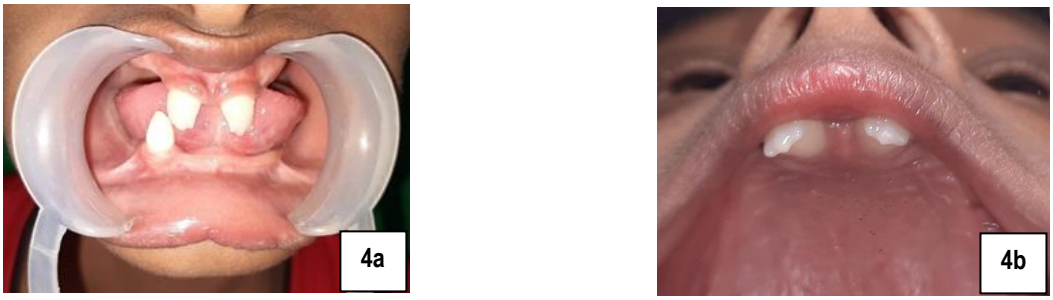

Figure 4 (4a, 4b)- Intraoral photographs of case 2

DISCUSSION

Terminology and Nomenclature: An Overview from Precedent to Neoteric Standing

Ectodermal Dysplasias are an outsized and intricate nosological assemblage of diseases, originally described by Thurnam in 1848. The term "hereditary ectodermal dysplasia", used as a surrogate for "dystrophy of the teeth and nails" or "congenital ectodermal defect", was first proposed by Weech. The disease was characterized into anhidrotic and hidrotic types by Clouston.[10] Currently, the term ectodermal dysplasia is too expansive and is not unmistakably reproducible. In accordance to the development fields by Spemann, acknowledging the multifarious overlapping network of the chief genes that are responsible for the development of ectoderm, the term ectodermal dysplasia reallocates to a more comprehensive terminology of "genodermatosis".[11]

Pathophysiognomy with Genomic Contribution

The pathogenesis of a wide spectrum of ectodermal dysplasias relies upon the phenomenon of embryogenesis which occurs in discrete tissue organizational fields with explicit communications among the germ layers. Distinct functional pathways have been

implicated in disease recognition and manifestation including Hedgehog signaling pathway, Wingless signaling pathway, TNF alfa signaling pathway, NF KB signaling pathway, ED signaling pathway, EDA, EDAR and EDARADD genes, P63 signaling pathway, Gap-junctions—connexin pathway, Axin pathway and various pathway mutations.[12] TNF-like TNFRs signalling pathways/NF-kB regulation patterns are targeted in cases of major ectodermal involvement, abnormal immune response, and functional abnormalities of CNS. Gene regulators' expression is targeted in cases with major skeletal involvement and endocrine deficits. Molecules for cytoskeleton maintainence and stability like connexins are pivotal in cases with hyperkeratosis, keratoderma, deafness, corneal anomalies and retinal degeneration.[13] The various forms of inheritance include autosomal dominant, autosomal recessive, X-linked dominant, X-linked semidominant, heterogenous and unidentified patterns.[10]

Classification Schemes

Categorization into subgroups is in accordance to the occurrence or nonoccurence of the four primary ectodermal dysplasia (ED) defects namely Trichodysplasia (ED1), Dental dysplasia (ED2), Onychodysplasia (ED3) and Dyshidrosis (ED4).[14] Relying on the aforesaid, there are about 150 different types of ectodermal dysplasias catalogued into one of the following subgroups made up from the primary ED defects: 1-2-3-4 (Rapp-Hodgkin hypohidrotic ED, Anhidrotic X-linked ED), 1-2-3 (Trichodento-osseous syndrome, Ellis-van Creveld syndrome), 1-2 (Gorlin's syndrome, Oculodentodigital syndrome), 1-3 (Palmoplantar hyperkeratosis and alopecia), 1-4 (Congenital ED of the face), 2-3-4 (Hypoplastic enamel-onycholysis-hypohidrosis), 2-3 (Nail dystrophy-deafness syndrome), 2-4 (Marshall's ED with ocular and hearing defects), 1-3-4 (Freire-Maia syndrome), 1-2-4, 3 and 4.[10,14] Also, ectodermal dysplasias can be incorporated into one of the following groups: Group 1 with defects in developemental regulation/epithelial-mesenchymal interaction and Group 2 incorporating diseases with cytoskeleton maintenance and cell stability disorders.[13]

Table 1: Comprehensive overview and comparative matrix of clinical presentations of ED

Clinical manifestation	Case 1	Case 2
Age	25 years	12 years
Gender	Male	Male
Ethnicity	Indian	Indian
Family history	Unyielding	Yielding
Temperature intolerance	✓	✓
Unexplained pyrexia	✓	✓
Stature	Short	Normal
Intelligence	Normal	Normal
Dry scaling skin	✓	✓
Atopic eczema	✓	No
Pigmentation	Perioral hyperpigmentation	Perioral hyperpigmentation
Wrinkles	✓	✓
Chronic rhinitis/pharyngitis	✓	No
Hoarseness of voice	✓	No
Dysphagia	✓	No
Reduced sweating	✓	✓
Reduced salivation	✓	✓
Reduced lacrimation	✓	✓
Hypotrichosis	✓	✓
Texture of scalp hair	Sparse, fine	Sparse, fine, brittle
Eyebrows	Absent	Sparse
Eyelashes	Absent	Sparse
Pubic and axillary hair	Sparse	Sparse
Facial dysmorphic features	Prominent supraorbital ridges, depressed nasal bridge, frontal bossing, low set ears, midfacial hypoplasia	Prominent supraorbital ridges, depressed nasal bridge, low set ears, midfacial hypoplasia
Hearing	Normal	Normal
Corneal dysplasia/cataract/strabismus	Absent	Absent
Photophobia	✓	No
Protuberant Lips	✓	✓
Pseudorhagades	✓	✓
Nasal bridge	Saddle shaped	Depressed
Dystrophic nails/leukonychia/ malformation/ onychodysplasia	Absent	Absent
Hypodontia/anodontia/oligodontia	✓	✓
Truncated/cone shaped teeth	✓	✓
caries prone	No	No
Dentition affected	Deciduous and permanent	Deciduous and permanent
Vertical dimension	Decreased	Decreased
Oro-Facial clefts	Absent	Absent
Palatal arch	Narrow and high	Relatively flat
Missing digits	No	No

Myriad of Clinical Manifestations

The various clinical findings in a case of suspected ectodermal dysplasia have been comprehensively overviewed in Table 1. The table also dispenses a comparative matrix of clinical presentations of the two cases discussed in this report.

Contemporary Deliberations and Clinical Diagnosis

The definition of ED has a subjective facet with its interpretation varying from author to author. The various schools of thought in light of describing ED include: (a) a singular disease (CST syndrome), (b) A binary etiology with two well-defined and clearly characterized diseases (CST and Clouston syndromes), each of them without any internal etiological heterogeneity, (c) A group of two "main" diseases, the anhidrotic and the hidrotic forms, with some heterogeneity, (d) A group of a few diseases (CST, Clouston, ectrodactyly-ectodermal dysplasia cleft lip and palate, Ellis-van Creveld syndrome and Congenital ectodermal dysplasia of the face) and (e) A large group of diseases (Hallerman- Streiff's, Oral-facial-digital associations of defects, Osteochondro-dysplasias, Goltz-Gorlin's, Hamartoses, etc.).[15] The various clinical tests that can be used the diagnosis of ectodermal Dysplasia include sweat pore count with yellow starch iodine, mosaic pattern of Blashko's lines, and chorionic villus sampling for prenatal diagnosis, Genetic testing for HED, X-linked recessive and autosomal dominant HED, EEC syndrome and AEC syndrome is available through GeneDx.[16] The diagnosis of ED, however, is multifarious and interdisciplinary with significant contribution of skin histopathology and sequence analysis of implicated genes to corroborate clinical diagnosis.

CONCLUSION

In these cases, early diagnosis, prompt treatment and referral for rehabilitation are the keys to boost the self-esteem and restore the normal functions of the oral cavity. Hence the dentists should have a thorough knowledge of the diagnosis of such cases.

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