ECTODERMAL DYSPLASIA: CASE REPORT & LITERATURE REVIEW

Deepak passi, Gagan Mehta, kuldeep vishwakerma, pranshu singh

Department of Oral and Maxillofacial Surgery, Faculty of Dental Sciences, King George's Medical University, Lucknow, India, corresponding author: A-122/5A, Shalimar garden, extension 2, Sahibabad, Ghaziabad, U. P., India, PIN-201005, Email - drdeepakpassi@gmail.com, Ph - 9717299524

ABSTRACT:

Ectodermal dysplasia is an extremely rare hereditary disease characterized by a congenital dysplasia of one or more ectodermal structures and their accessory appendages. Common manifestations include defective hair follicles and eyebrows, frontal bossing with prominent supraorbital ridges, nasal bridge depression, and protuberant lips. Intraorally, common findings are anodontia or hypodontia, conical teeth, and, consequently, generalized spacing. The patient may suffer from dry skin, hyperthermia, and unexplained high fever as a result of the deficiency of sweat glands. The present review focuses on the clinical manifestations, classifications, and diagnosis of ectodermal dysplasia. Here we present a case report of a 22-year-old male, exhibited many of the manifestations of ectodermal dysplasia.

KEYWORDS: Ectodermal dysplasia’s, Christ-Siemens-Touraine syndrome, Clouston syndrome, onychodysplasias, O.P.G (ortho pentomo gram)

INTRODUCTION:

The ectodermal dysplasia’s is an extremely rare and large, heterogeneous group of genetically inherited disorders that are characterised 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] the term ectodermal dysplasia was not coined until 1929 by Weech.[2]

The ectodermal dysplasias are congenital, diffuse, and nonprogressive. To date, more than 192 distinct disorders have been described. The most common ectodermal dysplasias are X-linked recessive hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome), and hidrotic ectodermal dysplasia (Clouston syndrome). Current classification of ectodermal dysplasias is based on clinical features. Pure ectodermal dysplasias are manifested by defects in ectodermal structures alone, while ectodermal dysplasia syndromes are defined by the combination of ectodermal defects in association with other anomalies.

Freire-Maia and Pinheiro proposed the first classification system of the ectodermal dysplasias in 1982, [3] with additional updates in 1994 and 2001. [4,5] Their original classification system stratified the ectodermal dysplasias into different subgroups according to the presence or absence of (1) hair anomalies or trichodysplasias, (2) dental abnormalities, (3) nail abnormalities or onychodysplasias, and (4) eccrine gland dysfunction or dyshidrosis.

Overall, the ectodermal dysplasias were classified into either group A disorders, which were manifested by defects in at least 2 of the 4 classic ectodermal structures as defined above, with or without other defects, and group B disorders, which were manifested by a defect in one classic ectodermal structure (1-4 from above) in combination with (5) a defect in one other ectodermal structure (ie, ears, lips, dermatoglyphics). Eleven group A subgroups were defined, each with a distinct combination of 2 or more ectodermal defects (eg, 2-4, 1-2-3, 1-2-3-4 from above). The group B disorders were indicated as 1-5, 2-5, 3-5, or 4-5 (from above).

With the recent identification of the causative genetic defect for a number of the ectodermal dysplasias, newer classification systems have been devised. In 2003, Lamartine reclassified the ectodermal dysplasias into the following 4 functional groups based on the underlying pathophysiologic defect: (1) cell-to-cell communication and signaling, (2) adhesion, (3) development, and (4) other.[6] Similarly, in 2001, Priolo and Laganà reclassified the ectodermal dysplasias into 2 main functional groups: (1) defects in developmental regulation/epithelial-mesenchymal interaction and (2) defects in cytoskeleton maintenance and cell stability.[7] Other classification systems categorize the
ectodermal dysplasias based on defects in cell-cell communication and signalling, adhesion, transcription regulation, or development.\[8\] Several ectodermal dysplasia syndromes may manifest in association with midfacial defects, mainly cleft lip, cleft palate, or both. The three most commonly recognized entities are (1) ectodermal dysplasia, ectrodactyly, and clefting (EEC) syndrome [9]; (2) Hay-Wells syndrome or ankyloblepharon, ectodermal dysplasia, and cleft lip/palate (AEC) syndrome; and (3) Rapp-Hodgkin syndrome. Ectodermal dysplasias is usually transmitted as an X linked recessive trait in which gene is carried by female and manifest in male [10]. The prevalence of population has been assessed as between 1:10000 and 1:100000 live male birth [11].

**Case report:** A 22 year old young male, presented to our department of oral maxillofacial surgery, king’s georges medical university, lucknow, U.P. for evaluation of teeth abnormalities. He had a history of delayed teething and recurrent high fevers during infancy. Patient’s mother reported that he is not able to sweat and is heat intolerance they had to apply some precautions to protect him from overheating during warm weather and physical exertion like wearing of wet clothes. On extra oral examination there was broad forehead with frontal prominence, high hair line, low seated ears, thick slightly everted lips with wrinkled skin periorbitally giving ‘old man look’ and perioral hyperpigmentation were evident (Figure (1a) (1b) (1c)). Scalp and body Hair, eyebrows and lashes are fine sparse and light in colour (Figure (2a, 2b)). The patient’s skin was scaly, smooth, dry, wrinkled and hypopigmented (Figure (4)). Nails are thin, light and slightly cracked (Figure (3)). Intraorally there is oligodontia, only six teeth were present that to in maxillary arch, i.e two small 1st molars, two central incisors and two peg shaped lateral incisors [13]. palate is shallow. Mandibular arch is completely edentulous and is resorbed (Figure (5a, 5b)) [14] and is denture wearer. Patient was investigated with Xray O.P.G and Lateral skull view showing class III skeletal tendency and reduced lower facial height with bilateral impacted mandibular canine (Figure (6, 7)). Other family member (first degree cousin) was also affected by mild degree of hypohidrosis and dental abnormalities. On the basis of clinical presentation, radiographic and family history evaluation, patient was diagnosed as a case of Ectodermal dysplasia.

**Profiles views:**

![Profile view showing Frontal prominence, high hair line, low seated ears, thick slightly everted lips with wrinkled skin periorbitally giving ‘old man look’ and perioral hyperpigmentation](image1.png)

![Scalp hair, eyebrows and lashes are fine, sparse and light in colour](image2.png)
Figure (3) Thin, light and slightly cracked nails

Intraoral views:

Figure (4) Scaly, smooth, dry, wrinkled, hypopigmented skin

Radiographs:

Figure (5a) Hypodontia with conical teeth

Figure (5b) Complete edentulous mandibular arch

Figure (5a) Hypodontia with conical teeth

Fig. 6 O.P.G showing hypodontia, resorbed facial height

Mandibular alveolar ridge with impacted canines

Fig. 7 class III skeletal tendency and reduced lower facial height

DISCUSSION:

Ectodermal dysplasias are described as "heritable conditions in which there are abnormalities of two or more ectodermal structures such as the hair, teeth, nails, sweat glands, cranial-facial structure, digits and other parts of the body. Ectodermal dysplasia is not a single disorder, but a group of syndromes which is usually transmitted as an X linked recessive trait in which gene is carried by female and manifest in male. More than 150 different syndromes have been identified. Despite some of the syndromes having different genetic causes the symptoms are sometimes very similar, diagnosis is usually by clinical observation often with the assistance of family medical histories. Three cardinal clinical features are (a) **Hypotrichosis** (sparseness of scalp and body hair). (b) **Hypohidrosis** (reduced ability to sweat). (c) **Hypodontia** (congenital absence of teeth). Others features may be Periorbital hyperpigmentation, depressed nasal bridge (saddle nose deformity),

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Decreased sebaceous secretions, nasal secretions, Lack of dermal ridges, underdevelopment of the alveolar ridge, Raspy voice, Fragile-appearing skin, Retruded appearance of the midface . Physical growth and psychomotor development are otherwise within normal limits.

Management of affected individuals targets the three cardinal features and is directed at optimizing psychosocial development, establishing optimal oral function, and preventing hyperthermia.

Wigs or special hair care formulas and techniques to manage sparse, dry hair. During hot weather, affected individuals must have access to an adequate supply of water and a cool environment, which may mean air conditioning, a wet T-shirt, and/or a spray bottle of water. Some individuals may benefit from "cooling vests". Affected individuals learn to control their exposure to heat and to minimize its consequences. Dental treatment, ranging from simple restorations to dentures, must begin at an early age. Bonding of conical shaped teeth in young affected individuals improves esthetics and chewing ability. Dental implants in the anterior portion of the mandibular arch have proven successful only in children over age seven years [12]

**CONCLUSION:**
Patients with ectodermal dysplasia are unique and oral problems must be evaluated individually to provide most idle treatment .They should be managed by team of paedriactician, prosthodontist, dermatologist, otolaryngologist, speech therapist and psychologist. Cosmic and prosthodontics measured should be instituted as early as possible, however success of any prosthesis depend on the cooperation and communication between the dental team and patient.

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