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HYPOHIDROTIC ECTODERMAL DYSPLASIA: A CASE REPORT AND LITERATURE REVIEW

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ABSTRACT: Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. Hypohidrotic ectodermal dysplasia is a congenital syndrome, characterized by hypotrichosis (hair is sparse, fine and weak; anomalies in the skin and nails), hypohidrosis (due to the paucity of sweat glands which in turn gives rise to sweat disorders) and hypodontia (partial, and occasionally total, absence of primary and/or permanent dentition). Most patients with EDA have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy, if unrecognized. Thus an early diagnosis is important. A case report of ectodermal dysplasia has been reported with review of literature.

KEY WORDS: Ectodermal dysplasias, hypotrichosis, hypohidrosis

INTRODUCTION: The ectodermal dysplasia is a heterogeneous group of inherited disorders which can be defined as primary defects in the development of 2 or more tissues derived from embryonic ectoderm¹. It was first described by Thurnam in 1848 and was coined by Weech² in 1929. Freire-Maia and Pinheiro described numerous varieties of ectodermal dysplasia involving all possible Mendelian modes of inheritance^{2,3}. Hypohidrotic ectodermal dysplasia is the most common with an incidence of one in 17000 people worldwide⁴. Clinical spectrum of ectodermal dysplasia include manifestations in the hair (hypotrichosis, partial or total alopecia), nails (dystrophic, hypertrophic, or abnormally keratinized), teeth (enamel defects or absence), and sweat glands (hypoplastic or aplastic) are usually affected⁵.

This article reviews clinical signs associated with ectodermal dysplasia and a case report.

CASE REPORT: A 18 -year-old male reported to the department of Oral Medicine and Radiology (A.B. Shetty Memorial Institute of Dental Sciences, Mangalore, Karnataka, India) with a chief

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complaint of missing upper and lower teeth since birth. Patient gives no history of exfoliation of teeth but gives a history of delayed eruption of teeth (at the age of 2yrs only). Patient also complains of dry skin and absence of sweat in his skin. He is intolerable to withstand hot water and hot environment. Patient gives history of dry mouth and difficulty in swallowing. On general examination, the patient was well oriented with time, place and was cooperative. On examination extra orally patient had scanty hair in eyebrows and eyelashes (Fig 1). There was frontal bossing and depressed ala of nose (Fig 2). His skin appeared soft, dry in face, upper and lower limbs. Nails appear normal. Scanty eyebrows were seen. Areas of hyperpigmentation were seen around ala of nose (Fig 1). Intra orally patient was partially edentulous teeth present were 16,26 appears conical (Fig 3). Upper and lower alveolar ridges appear normal. His panoramic radiograph was relevant to the above findings (Fig 4). So with the above clinical findings we came to an diagnosis of Hypohidrotic ectodermal dysplasia.

DISCUSSION: Hereditary ectodermal dysplasia is the defective formation of one or more structures derived from ectoderm¹. The first description of ectodermal dysplasia was given by Thurnam in 1848 and the term was coined by Weech² in 1929. Its inheritance is typically a cross-linked recessive trait so that the frequency and severity of the condition is more pronounced in males than in females. Freire- Maia described as a developmental defect which at embryonic level affects the ectoderm and therefore the tissues and structures derived from it^{2,3}. Thus it affects the development of keratinocytes and cause aberrations in the hair, sebaceous glands, eccrine and apocrine glands, nails, teeth, lenses and conjunctiva of the eyes, anterior pituitary gland, nipples and the ears⁴.

Hypohidrotic form/Christ-Seimens- Tourian Syndrome & Hidrotic form/Clouston syndrome⁵ are the two main clinical variants of ectodermal dysplasia. Classic clinical presentation of Hypohidrotic form comprises the triad of hypohidrosis, hypotrichosis and hypodontia. Usually X-linked recessive inheritance is seen. Males are affected severely, while females show only minor defects. In the hidrotic form teeth, hair and nails are affected⁶. The sweat glands are usually spared. It is usually inherited as an autosomal dominant trait. Other inheritance modalities like autosomal recessive have also been reported⁷.

Hypohidrotic characterized by soft, thin and dry skin with partial or complete absence of sweat glands which is responsible for the inability to perspire and intolerance to heat and frequent hyperthermia. The sebaceous glands are also defective or absent^{6,7}. Palms and soles are hyperkeratotic; pseudorhagades are present around the eyes. "plastic wrap" appearance of the skin is characteristic of hypohidrotic type in the new born. In the hidrotic type, sweat glands develop partially; ducts may be formed but secretory coils are absent and there are reduced number of sweat pores¹. Scalp hair is often fine, stiff and short. Eyelashes and eyebrows are scanty and often missing⁸. The characteristic facial features are: frontal bossing, depressed nasal bridge, prominent supra orbital ridges, prominent and obliquely set ears, midface is depressed, the lower third of the face appears small due to lack of alveolar bone development, lips are protuberant^{3,8}. Oligodontia is the most characteristic feature of the oral cavity. The teeth that are present have abnormal crown form which is conical in shape⁹. Cases have been reported where both primary and permanent dentition were congenitally missing¹⁰.

CONCLUSION: In this report we highlighted on the extra oral and intraoral findings of Hypohidrotic ectodermal dysplasia.

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TABLE 1 - Differences between the hydrotic and hypohydrotic forms of ectodermal dysplasia^{9,10}.

	Hydrotic	Hypohydrotic
Mode of Inheritance	Most often autosomal dominant	Most often autosomal recessive
Scalp Hair	Soft, dawning, color is darker	Fine in texture, fair and short
Teeth	Anodontia to hypodontia	Anodontia to hypodontia
Lips	No abnormality	Protruding
Sweat glands	Active	Reduced to absent
Nasal bridge	No flattening	Underdeveloped
Nails	Dystrophic nails	No abnormality
Eyelashes/Pubic/Axillary hairs	Scanty/absent	Variably affected

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Figure 1- Clinical photograph showing scanty hair in eyebrows and eyelashes areas of hyperpigmentation were seen around ala of nose

Figure 2- frontal bossing and depressed ala of nose

Figure 3- . Intra orally patient was partially edentulous.teeth present were 16,26 appears conical

Figure 4- panoramic view