Hereditary ectodermal dysplasia: a case report

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Hereditary ectodermal dysplasia is an inherited disorder involving skin, hair, nails and teeth. Two main clinical forms have been described - hypohidrotic type and hidrotic type. A case of ectodermal dysplasia with absence of hypohidrosis and defective nails has been reported. The importance of early prosthetic management has been discussed.


KEY WORDS: Ectodermal dysplasia, Prosthetic management.

Ectodermal dysplasia is the term used for a group of inherited disorders involving skin, hair, nails and teeth. Freire-Maia and Pinheiro described 117 possible varieties of ectodermal dysplasia involving all possible Mendelian modes of inheritance. From the clinical point of view two main forms have been distinguished:


The Hypohidrotic form exhibits the classic triad - 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 affected. The sweat glands are usually spared. It is usually inherited as an autosomal dominant trait. Other inheritance modalities like autosomal recessive have also been reported.

In the hypohidrotic form, the skin is soft, thin and dry. Partial or complete absence of sweat glands is responsible for the inability to perspire and accounts for intolerance to heat and frequent hyperthermia. The sebaceous glands are also defective or absent. Palms and soles are hyperkeratotic, psuedorrhagades are present around the eyes. Atopic eczema is common, especially in flexures during early childhood. In a new born, a "plastic wrap" appearance of the skin is characteristic of hypohidrotic type. In the hidrotic type, sweat glands develop partially; ducts may be formed but secretory coils are absent and there are reduced number of sweat pores. Scalp hair is often fine, stiff and short. Eyelashes and eyebrows are scanty and often missing.

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. A cephalometric study by Vierucci and co-workers have shown significant differences in craniofacial features of unaffected and affected children. Children with hypohidrotic type ectodermal dysplasia showed maxillary retrusion due to sagitally underdeveloped maxilla, forward and upward displacement of the mandible and collapsed lower anterior facial height.

In the oral cavity, the most striking feature is oligodontia. The teeth that are present have abnormal crown form. Teeth in the anterior region of maxilla and mandible are conical in shape. There is a wide midline diastema and hypoplastic labial frenum. Cuspsids are usually normal. Commonly, there is only one molar tooth in the second molar region which usually exhibits a bud crown form. Consistent variations in the number and crown forms of teeth occur which appear to be a characteristic dental phenotype for ectodermal dysplasias with different modes of inheritance. This is demonstrated especially in autosomal recessive condition where there is a total absence of permanent teeth with or without taurodontism of primary molars. Cases have been reported where both primary and permanent dentition were congenitally missing.