

Hypohidrotic ectodermal dysplasia

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A 16-year-old female patient presented with the complaint of missing teeth. Examination revealed frontal bossing, depressed nasal bridge and sunken cheeks, xerotic skin, dry thin lusterless sparse hair over the scalp and eyelashes [Figures 1 and 2]. Lips were thick, full and everted [Figure 3]. Periorbital and perioral skin wrinkling with hyperpigmentation was noted. Oral mucosa, nails, palmoplantar surfaces were normal. Cone shaped 53, 63 with microdontic 24, 25 was observed [Figures 3 and 4]. The remaining alveolar ridges were completely edentulous [Figures 3 and 5]. The patient had hearing loss with near sighted vision, decreased salivation, complete absence of sweating and mental retardation.

Ectodermal dysplasias are a group of X-linked inherited disorders characterized by dysplasia of tissues of ectodermal origin with prevalence of approximately 1:1,00,000 live births and is

caused by genetic defects in ectodysplasin signal transduction pathways located at Xq12-13.^[1,2] It is usually seen in males and is inherited through female carriers.^[3] Hypohidrotic ectodermal dysplasia is characterized by three classical triad of hypodontia, hypotrichosis, and hypohidrosis. Dry scaly skin and unexplained pyrexia and heat intolerance occur due to anhidrosis due to their diminished ability to sweat.^[4,5]

Intraorally, multiple missing teeth especially permanent teeth, root, and crown dysmorphism (such as conical teeth) and reduced salivary flow is seen.^[2,4,5] Very rarely, one or both jaws may be edentulous.^[4] A depressed nasal bridge (saddle nose) gives the impression of a small sized face.^[5]

Treatment is symptomatic management that involves multidisciplinary approaches as oral rehabilitation, restorative procedures, and maintenance of cool, ambient temperature to prevent hyperthermia.^[1]

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Figure 1: Absent eyebrow hair, frontal bossing, saddle nose, everted lips, periorbital and perioral hyperpigmentation, low positioning of ears



Figure 2: Sparse hair distribution on scalp



Figure 3: Cone shaped deciduous canines, hypodontic maxilla, xerotic skin



Figure 5: Completely edentulous mandibular ridge



Figure 4: Normal palmoplantar surfaces

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