Hypohidrotic Ectodermal Dysplasia: Classical Clinical Features

A 9-year-old boy presented to the dermatology OPD with complaints of poor scalp hair growth, loss of sweating, heat intolerance and delayed eruption of teeth. Child was born uneventfully out of a nonconsanguineous marriage at term by vaginal route. He was first in birth order with the other two siblings being normal. Examination revealed frontal bossing, depressed nasal bridge, saddle nose deformity, marked periocular and perioral hyperpigmentation, prominent lips and ears (**Fig. 1**). Oral cavity examination showed only three widely spaced teeth (one upper molar and two lower molars). Scalp hair were sparse, thin and lustreless with scant eyelashes, absent eyebrows and body hair. The skin was overall dry, with reduced dermatoglyphics on palms and soles while nails were normal.

Hypohidrotic ectodermal dysplasia (HED), also known as Christ-Siemens-Touraine syndrome, presents with a triad of hypodontia, hypotrichosis and hypohidrosis. It has variable inheritance as X-linked, autosomal recessive or autosomal dominant. Distinctive cutaneous features include periocular and perioral hyperpigmentation, dry wrinkled skin, eczematous dermatitis and sebaceous hyperplasia. A mid-facial hypoplasia leads to the characteristic facies. The condition is associated with abnormal mucosal glands leading to solidified aural and nasal secretions, dry eyes and recurrent pulmonary infections. Management of HED is challenging, requiring a multidisciplinary approach including prevention of hyperthermia, managing eczematous dermatitis and recurrent pulmonary infections. Early dental intervention to improve the chewing ability improves the overall quality of life.

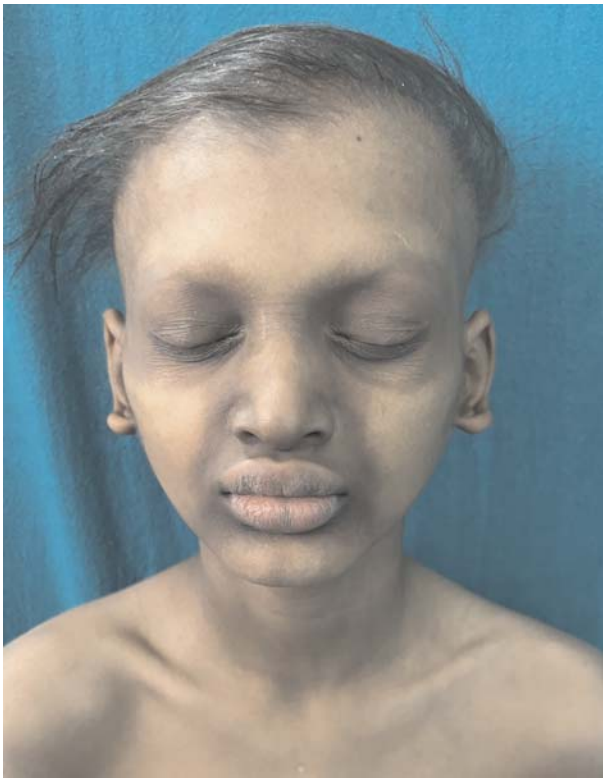

Fig. 1 Characteristic facies showing a broad forehead, sparse eyebrows, depressed nasal bridge, prominent lips, and periocular and perioral hyperpigmentation

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