

Hypohidrotic Ectodermal Dysplasia

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Fig. 1 : Dymorphic facies with frontal bossing, prominent supraorbital ridges, absence of eyebrow and eyelashes, and a saddle nose



Fig. 2 : Open mouth shows conical teeth and impression of loss of teeth

A 20-year-old unmarried girl was admitted with complaints of poor formation of saliva along with frequent dental caries, and poor formation of tear associated with recurrent conjunctivitis. She had moderate grade of intermittent fever for last few years often complicated by pharyngitis, rhinitis and otitis. There was history of loss of teeth from time to time with painful mastication. Past history revealed febrile convulsions in childhood, normal milestones of development except delayed eruption of permanent tooth, and there was absence of burning micturition/ cough/ discharging ears/ any sort of joint ailment/ or similar type of illness in family members. On examination, she had slightly poor physical but normal mental development. She had a dysmorphic face characterized by frontal bossing, prominent supraorbital ridges with lack of eyebrows and eyelashes, saddle nose with big nostrils, thick-everted lips with sunken cheeks, narrow lower part of face, wrinkled and hyperpigmented periorbital skin (Fig.1); there was absence of rhagades, perforated nasal septum and upper central incisors. Skin showed chronic eczematous dermatitis in places along with rise of temperature. Body hair was sparse, fine, brittle and lusterless; alopecia was prominent. Nails showed onychodysplasias, and teeth were conical and totally 11 in number (Fig.2). Other than tachycardia all other vital parameters and systemic examinations were essentially normal. R/E of her blood, urine, and chest X-ray were inconclusive. So, primarily a disease involving ectoderm (i.e., skin, hair, nails, teeth and eccrine glands) was suspected with a provisional diagnosis of ectodermal dysplasia (ED); other differential diagnoses considered were congenital syphilis, lepromatous leprosy, Wegener's granulomatosis, midline granuloma and Sjogren's syndrome. During her hospital stay, surprisingly, the patient disclosed that she likes swimming hours after hours in the local pond, favours to lie in the cement floor than in cot/bed, prefers indoor games than outdoor, experiences very high temperature even in winters, and habituated with lack of sweating. So, eccrine gland dysfunction or dyshidrosis were clinically documented.

The ectodermal dysplasias comprise a large heterogeneous group of inherited disorders which share primary defects in the development of two or more tissues derived from embryonic ectoderm. The term ectodermal dysplasia was coined by Weech in 1929. There are mainly two types EDs: hypohidrotic ED (Christ-Siemens-Touraine syndrome), and hidrotic ED (Clouston syndrome) though there are many subtypes. X-linked hypohidrotic ED is the most common variety. Female carriers may be affected as a result of lyonization and somatic mosaicism for the abnormal X-chromosome; autosomal recessive and autosomal dominant forms of hypohidrotic ED are very rare. The present case revealed skin abnormality, trichodysplasias, dental abnormalities, onychodysplasias, and dyshidrosis which are included under one subgroup of ED. Sweat pore counts (performed by using yellow starch-iodine powder), pilocarpine iontophoresis and skin biopsy (from hypothenar eminence) may document hypohidrosis, and a reduction in the number of eccrine glands. There is no definite treatment of ED and antipyretics are not effective in the management of pyrexia.

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