Letter to the Editor

Hypohydrotic ectodermal dysplasia: a case report

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Sir,

A 23 years old girl walked into our clinic with the complaint of sparse hairs over the scalp since childhood (Fig. 1). She revealed that consultation with various doctors from different streams could not be of any help to her till date. The scalp hairs were dull looking and lustreless and eyebrows were scanty with the outer third completely missing (Fig. 2). However, the eyelashes were normal. Hair in the axilla, pubic region and extremities were also sparse. The skin was dry looking. Further questioning revealed that she sweats very meagerly and was also intolerant to heat. However, she had never suffered stroke in the past. There were no scaling or peeling of skin noted. Periorbital hyperpigmentation with fine wrinkles around the eyes were evident. Nails were normal on examination. Her lower lips were slightly everted (Fig. 2). Oral examination found lower set of teeth completely missing. She did not have the mandibular teeth from birth. She had delayed dentition with only 8 maxillary deciduous teeth appearing which later got replaced with the same number of permanent teeth. She was a girl of normal intelligence. Her systemic examination was within normal limits. She had reached puberty on time with normal breast development and her menstrual cycles were regular. Her two siblings and parents were healthy without any similar complaints. A diagnosis of hypohydrotic ectodermal dysplasia (HED) was made based on clinical features.

Ectodermal dysplasia (ED) is defined as a large heterogeneous group of conditions, characterized by congenital defects in two or more ectodermal derivatives such as hair, teeth, nails or sweat glands. The classification of 2014 include 163 defined ED syndromes [1]. The most common EDs are X-linked recessive hypohydrotic ED (Christ Siemens Touraine Syndrome) (HED) and hydrotic ED (Clouston Syndrome). HED is characterized by the triad of hypotrichosis, hypodontia and hypohydrosis. Our case classically demonstrated this triad. It is inherited as an X-linked disorder; however autosomal dominant and recessive forms have been described [2]. Defect in three genes: Ectodysplasin A (EDA), EDA receptor (EDAR) and EDAR associated death domains (EDARADD) are implicated [3]. Females have generally partial manifestation of the disease unlike males who have more generalized features [4]. Beside the triad as described above, other clinical features may include nail disorders, craniofacial abnormalities, respiratory infections, eczema, and others. However, clinical manifestations are highly variable among individuals.

Hypohydrosis especially during infancy and childhood may cause thermoregulatory issues. In fact, in anhydrotic ectodermal dysplasia, heat stroke has been the most common cause of death in first year of life.

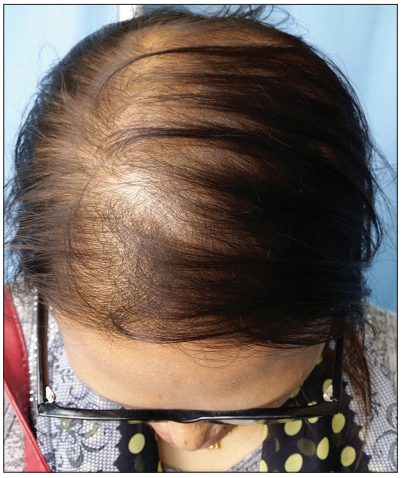

Figure 1: Sparse hairs over the scalp.

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Figure 2: Scanty eyebrows with the outer third missing, periorbital hyperpigmentation with fine wrinkles, and everted lips.

Thermal imbalance can be countered by drinking cold fluids, wetting the skin and preferring cooler place to live and work. The sparse hair is cosmetically unacceptable, more so among female patients. This was evident in our case as well. The alopecia is rarely total. Recently, topical application of 3% minoxidil for a year has achieved good results [5]. Delayed dentition is perhaps the first symptom that makes the parents seek medical advice. Dental issues range from anodontia to hypodontia, widely spaced to conical/pegged shaped teeth. Both temporary as well as permanent teeth are affected. My patient too did not have lower set of teeth. Immediate dental attention is warranted which is often challenging. A combined surgical, pedodontic and prosthodontic approach is prescribed. Nail changes could include onychodysplasia.

The diagnosis of HED may not be very difficult. However, it requires multidisciplinary approach to manage such patients. Counselling, prosthodontic treatment and dermatological consultation must be offered to every patient.

CONSENT

The examination of the patient was conducted according to the Declaration of Helsinki principles.

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