Hypohidrotic ectodermal dysplasia (HED) refers to a group of disorders that share the following features: sparse or absent hair; absent or peg-shaped teeth; and decreased ability to sweat. HED is a life-long disease therefore it’s essential early on to educate the parents on ways to prevent and control hyperpyrexia.

Physical examination revealed diffuse pallor with erythematous patches on bilateral cheeks and scattered impetiginized, excoriated papules and plaques on the philtrum, wrists, ankles and antecubital fossa with moderate lichenification. Examination of patient’s scalp revealed blonde hair with thick chalky adherent scale. Sparse eyebrows, minimal body hair and hypodontia were also observed.

We began treating patients impetiginized eczema with triamcinolone 0.1% cream and mupirocin 2% ointment. Bleach baths twice weekly, wet wraps and daily topical emollients were encouraged. A referral to genetics was encouraged for further work up of presumed anhidrotic ectodermal dysplasia as there is a strong family history of the disease. Handouts were provided, and we continue to follow the patient for the development of any additional disease manifestations.

Hypohidrotic ectodermal dysplasia (HED), also known as anhidrotic ectodermal dysplasia or Christ–Siemens–Touraine syndrome refers to a group of disorders that share the following features: sparse or absent hair; absent or peg-shaped teeth; and decreased ability to sweat. The most common form is X-linked inherited occurring in approximately 1 in 10,000 live-born males, but both autosomal dominant and autosomal recessive inheritance patterns have been documented. It is likely that our patient has the X-linked or autosomal dominant variant. HED affects the developing nail, hair follicle and eccrine gland through a genetic defect in ectodysplasin signal transduction pathway. In the X-linked form of HED ectodysplasin A, which is secreted by epithelial cells, is defective. In the autosomal dominant and recessive forms ectodysplasin-A receptor (EDAR) is the underlying defect. Signaling errors ultimately translate into the nucleus with the help of NF-κB and result in aplasia, hypotonia or dysplasia of these structures.

Clinically newborns present encased in a collodion-like membrane or with skin scaling. Scalp hair may be absent, sparse, or when present is typically blonde. Body hair is sparse to absent. Newborns are unable to sweat and often present with pyrexia of unknown origin. Eczema, periorbital wrinkling, and hyperpigmentation are common. Nails are usually unaffected, but hypodontia, anodontia, and/or conical teeth are common. Patients may also have saddle nose, everted lips and frontal bossing. Female patients with the X-linked form have variable involvement due to the random nature of X-inactivation presentations.

HED is a life-long disease therefore it’s essential early on to educate the parents on ways to prevent and control hyperpyrexia.

Multidisciplinary care is often required for the treatment of upper respiratory symptoms, dental complications, and atopy. Referral to the National Foundation for Ectodermal Dysplasias is also an important aspect of care. Gene and protein therapy for HED is on the horizon.