Hypohidrotic ectodermal dysplasia

Genetics

-EDA (Ectodysplasin A), EDAR, EDARADD

-Mostly XLR (EDA:95%), AD or AR (5%)

Clinical findings/Dysmorphic features

-Peeling skin and perioral hyperpigmentation at birth

-Hypotrichosis (sparseness of scalp and body hair)

-Hypohidrosis (reduced ability to sweat)

-Hypodontia (congenital absence of teeth)

Etiology

-1 in 5,000 – 1 in 10,000

Pathogenesis

-Defective ectodysplasin A cannot be activated to mediate the cell-to-cell signaling that regulates morphogenesis of ectoderm

Genetic testing/diagnosis

-Diagnosed after infancy on the basis of physical features in most affected individuals

-Hemizygous EDA pathogenic variant in an affected male

-Biallelic EDAR, EDARADD, or WNT10A pathogenic variants in affected male or female

-EDA sequencing (~95% XL HED), EDAR and EDARADD sequencing

Others

-Wigs and saliva substitutes