Hidrotic ectodermal dysplasia 2

Genetics

-GJB6 (13q12) only known gene

-AD

Clinical findings/Dysmorphic features

-Malformed, thickened, small nails

-Hypotrichosis (partial or total alopecia)

-Palmoplantar hyperkeratosis

-Normal sweating and normal teeth

Etiology

-Common in the French-Canadian population of southwest Quebec

Pathogenesis

-Helps to form a gap junction channel which mediates ion diffusion

-Mutations affect trafficking of the protein and thus the formation of the gap junction

Genetic testing/diagnosis

-Targeted analysis for the four known pathogenic variants in GJB6 (p.Gly11Arg, p.Val37Glu, p.Asp50Asn, p.Ala88Val); account for 100%