Hypohidrotic ectodermal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypohidrotic ectodermal dysplasia. ORPHA:238468

Hypohidrotic ectodermal dysplasia (HED) is a genetic disorder of ectoderm development characterized by malformation of ectodermal structures such as skin, hair, teeth and sweat glands. It comprises three clinically almost indistinguishable subtypes with impaired sweating as the key symptom: Christ-Siemens-Touraine (CST) syndrome (X-linked), autosomal recessive (AR), and autosomal dominant (AD) HED, as well as a fourth rare subtype with immunodeficiency as the key symptom (HED with immunodeficiency) (see these terms).