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# Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base.*

*Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome. ORPHA:363523*

Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by severe intellectual disability with significant speech and language impairment, hypohidrosis (often resulting in hyperthermia) with normal sweat gland appearance, tooth enamel hypoplasia, palmoplantar hyperkeratosis and a high frequency of acquired microcephaly. Mild facial dysmorphism, including lateral flaring of the eyebrows, broad nasal tip, and thick vermilion border, may also be observed.