

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

Shaheen syndrome

Shaheen syndrome

Kod ORPHA

363523

Kod OMIM

615328

Kod ICD10

Q82.8

Kod ICD11

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*Źródło

orphanet