

Hydrotyczna dysplazja ektodermalna typu Halal

Kod Orpha: 1809 Kod OMIM:

Opis choroby *

Definicja

Hidrotic ectodermal dysplasia, Halal type is a form of ectodermal dysplasia syndrome (see this term) characterized by trichodysplasia, with absent eyebrows and eyelashes, onychodysplasia, mild retrognathia, abnormal dermatoglyphics (excess of whorls on fingertips, radial loop on finger, hypothenar pattern), intellectual disability and normal teeth and sweating. Additional variable manifestations include high implanted or prominent ears, mild hearing loss, supernumerary nipple, café-au-lait spots, keratosis pilaris, and irregular menses. To date, four individuals from 2 generations of a consanguineous family of Portuguese descent have been described in the literature. Males and females were equally affected. Hidrotic ectodermal dysplasia, Halal type is inherited in an autosomal recessive manner.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Halal-Setton-Wang syndrome
Zespół Halal, Setton i Wanga
Trichodysplasia-abnormal dermatoglyphics-intellectual disability syndrome

Kod ORPHA

1809

Kod OMIM

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Kod ICD10

Q82.8

Kod ICD11

LD27.0Y

[*Źródło](#)

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Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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