

Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by signs of ectodermal dysplasia (sparse hair, abnormal or missing teeth, decrease or absent sudation), typical facial features (protruding forehead, wrinkles under the eyes, characteristic periorbital hyperpigmentation), and immunodeficiency.

Dane

Klasyfikacja

Choroba

Synonimy

Anhidrotic ectodermal dysplasia with immunodeficiency

Anhidrotyczna dysplazja ektodermalna z niedoborem odporności

EDA-ID

EDA-ID

HED-ID

Kod ORPHA

98813

Kod OMIM

612132

Kod ICD10

D82.8

Kod ICD11

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*Źródło

orphanet