

Opis choroby *

Definicja

A rare genetic disease characterized by progressive and severe sensorineural hearing loss with onset in the first decade of life, associated with mild thrombocytopenia, often with enlarged platelets. Most patients do not show significant bleeding tendency.

Dane

Klasyfikacja

Choroba

Synonimy

DIAPH1-related sensorineural deafness-thrombocytopenia syndrome

DIAPH1-related sensorineural deafness-thrombocytopenia syndrome

Kod ORPHA

494444

Kod OMIM

124900

Kod ICD10

H90.3

Kod ICD11

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*Źródło

orphanet