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