

Opis choroby *

Definicja

A rare, genetic, isolated constitutional thrombocytopenia disease characterized by decreased platelet counts, not associated with platelet morphology or function impairment, in multiple members of a family. Manifestations are variable, typically ranging from asymptomatic to mild bleeding diathesis (e.g. easy bruising, epistaxis, petechiae). Occasionally, a more severe bleeding tendency has been associated and a mild predisposition to infection and eczema has been reported.

Dane

Klasyfikacja

Choroba

Kod ORPHA

268322

Kod OMIM

612004

Kod ICD10

D69.4

Kod ICD11

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*Źródło

orphanet