Opis choroby *

Definicja

A rare isolated hereditary giant platelet disorder characterized by severe thrombocytopenia and thrombopathy due to defects in proplatelet formation and platelet activation in homozygous patients. Clinical manifestation are recurrent bleeding episodes including epistaxis, spontaneous hematomas, and menorrhagia.

Dane

Klasyfikacja
Choroba

Kod ORPHA
438207

Kod OMIM
616176

Kod ICD10
D69.4

Kod ICD11
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*Źródło

orphanet