Opis choroby *

Definicja

A rare genetic hematologic and intestinal disease characterized by childhood onset of bleeding tendency with epistaxis, gum bleeding, gastrointestinal bleeding, hematuria, and menorrhagia due to impaired platelet aggregation and secretion, as well as recurrent gastrointestinal ulcera. Mildly reduced levels of coagulation factor XI have been reported in addition.

Dane

Klasyfikacja
Choroba

Synonimy
PLA2G4A-related platelet dysfunction
Dysfunkcja płytek spowodowana niedoborem cytozolowej fosfolipazy A2 alfa
Dysfunkcja płytek związana z plA2G4A
Platelet dysfunction due to cytosolic phospholipase-A2 alpha deficiency

Kod ORPHA
477787

Kod OMIM
618372

Kod ICD10
D69.1

Kod ICD11
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*Źródło

orphanet