

## 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 ulcers. 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

-

---

#### \*Źródło

orphanet