

Opis choroby *

Definicja

Bleeding disorder due to CalDAG-GEFI deficiency is a rare hematologic disease due to defective platelet function and characterized by mucocutaneous bleeding starting in infancy (around 18 months of age), presenting with prolonged and severe epistaxis, hematomas and bleeding after tooth extraction. Massive menorrhagia and chronic anemia have also been reported.

Dane

Klasyfikacja

Choroba

Synonimy

Bleeding disorder due to calcium- and DAG-regulated guanine exchange factor-1 deficiency
Skaza krwotoczna z powodu niedoboru czynnika wymiany nukleotydów guaninowych-1 regulowanego wapniem i DAG

Kod ORPHA

420566

Kod OMIM

615888

Kod ICD10

D69.1

Kod ICD11

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*Źródło

orphanet