Opis choroby *

Definicja

A rare genetic coagulation disorder characterized by marked bleeding tendency and posttraumatic bleeding with easy bruising, soft tissue and muscle bleeding, hemarthroses, and menorrhagia due to an increase of soluble thrombomodulin in plasma with subsequent protein C activation and reduction of thrombin generation within a potential thrombus. Abnormal laboratory findings include markedly elevated plasma thrombomodulin, reduced prothrombin consumption, and decreased thrombin generation.

Dane

Klasyfikacja
Choroba

Synonimy
THBD-related bleeding disorder
Koagulopatia zależna od THBD
Koagulopatia zależna od trombomoduliny
Zaburzenia krzepnięcia zależne od THBD
THBD-related coagulopathy
Thrombomodulin-related coagulopathy

Kod ORPHA
436169

Kod OMIM
614486

Kod ICD10
D68.3

Kod ICD11
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*Źródło

orphanet