DISEASE:
Severe autosomal recessive macrothrombocytopenia

NAME:	Severe autosomal recessive macrothrombocytopenia
DESCRIPTION:	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.
ORPHACODE:	438207
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PRKACG GNE
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RELATED CONTENT

Related Genetic Tests

- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase
- protein kinase cAMP-activated catalytic subunit gamma

Related Gene Panels

- Trombosis - Hemostasis (107 genes) - KUL