DISEASE:
Autosomal dominant thrombocytopenia with platelet secretion defect

NAME:	Autosomal dominant thrombocytopenia with platelet secretion defect
DESCRIPTION:	A rare isolated constitutional thrombocytopenia characterized by reduced platelet count and defective platelet ATP secretion, resulting in increased bleeding tendency. Clinical manifestations are easy bruising, gum bleeding, menorrhagia, spontaneous epistaxis, spontaneous muscle hematoma, and potential postpartum hemorrhage, among others.
ORPHACODE:	466806
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	SLFN14
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