
DISEASE:
Thrombocythemia with distal limb defects

NAME:	Thrombocythemia with distal limb defects
DESCRIPTION:	Thrombocythemia with distal limb defects is a rare, genetic syndrome with limb reduction defects characterized by thrombocytosis, unilateral transverse limb defects (ranging from absence of phalanges to absence of hand or forearm) and splenomegaly.
ORPHACODE:	329319
SYNONYMS:	Familial thrombocytosis with transverse limb defect Hereditary thrombocytosis with transverse limb defect
XREF(S):	Orphanet
ANALYTE(S):	THPO
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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