Hereditary thrombocytopenia with normal platelets

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hereditary thrombocytopenia with normal platelets. ORPHA:268322

A rare, genetic, isolated constitutional thrombocytopenia disease characterized by decreased platelet counts, not associated with platelet morphology or function impairment, in multiple members of a family. Manifestations are variable, typically ranging from asymptomatic to mild bleeding diathesis (e.g. easy bruising, epistaxis, petechiae). Occasionally, a more severe bleeding tendency has been associated and a mild predisposition to infection and eczema has been reported.