

DISEASE:
Autosomal dominant macrothrombocytopenia

NAME:	Autosomal dominant macrothrombocytopenia
DESCRIPTION:	A rare isolated constitutional thrombocytopenia characterized by abnormally large platelets.
ORPHACODE:	140957
XREF(S):	Orphanet OMIM OMIM ICD-10 OMIM OMIM OMIM

ANALYTE(S):	<u>TUBA8</u> <u>GP1BA</u> <u>TRPM7</u> <u>GFI1B</u> <u>GP1BB</u> <u>TPM4</u> <u>ITGA2B</u> <u>ITGB3</u> <u>TUBB1</u> <u>ACTN1</u>
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RELATED CONTENT

Related Genetic Tests

- [Coagulopathies \(2 genes\)](#)
- [Trombosis - Hemostasis \(gene panel\)](#)

Related Laboratories

- [Centre de Génétique Médicale UCL](#)
- [Centrum Menselijke Erfelijkheid - KUL](#)

Related Analytes

- [actinin alpha 1](#)
- [growth factor independent 1B transcriptional repressor](#)
- [glycoprotein Ib platelet subunit alpha](#)
- [glycoprotein Ib platelet subunit beta](#)
- [integrin subunit alpha 2b](#)
- [integrin subunit beta 3](#)
- [tropomyosin 4](#)
- [transient receptor potential cation channel subfamily M member 7](#)
- [tubulin alpha 8](#)
- [tubulin beta 1 class VI](#)

Related Gene Panels

- Coagulopathie (2 genes) - UCL
- Trombosis - Hemostasis (107 genes) - KUL

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