

Thrombocytopenia / Thrombocytopathy

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
Bernard Soulier-Syndrome (BSS) ⁺	<i>GP1BA</i> (CDS complete) <i>GP1BB</i> (CDS complete) <i>GP9</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Familial platelet disorder with predisposition to AML ⁺	<i>GATA1</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Hermansky Pudlak - Syndrome ⁺	<i>AP3B1</i> (CDS complete) <i>BLOC1S3</i> (CDS complete) <i>DTNBP1</i> (CDS complete) <i>HPS1</i> (CDS complete) <i>HPS3</i> (CDS complete) <i>HPS4</i> (CDS complete) <i>HPS5</i> (CDS complete) <i>HPS6</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Glanzmann thrombasthenia (GT) ⁺	<i>ITGA2B</i> (CDS complete) <i>ITGB3</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
MYH9-related diseases (May-Hegglin-Anomaly (MHA), Sebastian-Syndrome (SBS), Epstein-Syndrome (EPS), Fechtner-Syndrome (FTNS)) ⁺	<i>MYH9</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Storage Pool Disease - alpha ⁺	<i>NBEAL2</i> (CDS complete) <i>PLAU</i> (CDS complete) <i>VPS33B</i> (CDS complete) <i>VIPAS39</i> (CDS complete) <i>P2RY12</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS

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Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
Storage Pool Disease - delta ⁺	<i>AP3B1</i> (CDS complete) <i>BLOC1S3</i> (CDS complete) <i>DTNBP1</i> (CDS complete) <i>HPS1</i> (CDS complete) <i>HPS3</i> (CDS complete) <i>HPS4</i> (CDS complete) <i>HPS5</i> (CDS complete) <i>HPS6</i> (CDS complete) <i>NBEA</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Storage Pool Disease - alphadelta ⁺	<i>WAS</i> (CDS complete) <i>GATA1</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Thrombozytopenia with large platelets ⁺	<i>GATA1</i> (CDS complete) <i>MYH9</i> (CDS complete) <i>FLI1</i> (CDS complete) <i>NBEAL2</i> (CDS complete) <i>GP1BA</i> (CDS complete) <i>GP1BB</i> (CDS complete) <i>GP9</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Thrombozytopenia with normal platelets ⁺	<i>GATA1</i> (CDS complete) <i>RUNX1</i> (CDS complete) <i>RBM8A</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Thrombozytopenia with small platelets ⁺ , Wiskott-Aldrich-Syndrom ⁺	<i>WAS</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS

Note:

According to the Genetic Diagnostics Act, every examination request for human genetic diagnostics must be accompanied by a declaration of consent from the patient or his legal representative.

Forms for this as well as request forms for the analyses are available in the download area of our homepage.