

Coagulation Factor- / Platelet- / Thrombotic-Disorders

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
ADAMTS13 deficiency ⁺	<i>ADAMTS13</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Antithrombin deficiency	<i>SERPINC1</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
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
Prothrombin deficiency ⁺	<i>F2</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Factor V deficiency ⁺	<i>F5</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Factor VII deficiency ⁺	<i>F7</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
Hemophilia A ⁺	<i>F8</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	PCR (Inversion Intron 1 and Intron 22) NGS MLPA
Hemophilia B ⁺	<i>F9</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
Factor X deficiency ⁺	<i>F10</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
Factor XI deficiency ⁺	<i>F11</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS

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Factor XII deficiency ⁺	<i>F12</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Factor XIII deficiency ⁺	<i>F13A1</i> (CDS complete) <i>F13B</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Fibrinogen deficiency ⁺	<i>FGA</i> (CDS complete) <i>FGB</i> (CDS complete) <i>FGG</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Fletcher factor deficiency ⁺	<i>KLKB1</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Kininogen deficiency ⁺	<i>KNG1</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Glanzmann thrombasthenia ⁺	<i>ITGA2B</i> (CDS complete) <i>ITGB3</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Hereditary angioedema type 1 & 2 ⁺	<i>SERPING1</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Hereditary angioedema type 3 ⁺	<i>F12</i> (Exon 9)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Hereditary hemorrhagic telangiectasia ⁺	<i>ACVRL1</i> (CDS complete) <i>ENG</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
Plasminogen deficiency ⁺	<i>PLG</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS

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Protein C deficiency	<i>PROC</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
Protein S deficiency	<i>PROS1</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
Von Willebrand disease	<i>VWF</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS MLPA
Wiskott-Aldrich syndrome ⁺	<i>WAS</i> (CDS complete)	5 ml EDTA blood	as needed 2-6 weeks	NGS
Antithrombin III Cambridge II A384S (rs121909548)	<i>SERPINC1</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
Prothrombin G19911A (rs3136516)	<i>F2</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
Prothrombin G20210A (rs1799963)	<i>F2</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
Factor V Leiden R506Q (rs6025)	<i>F5</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
Factor V H1299R (rs1800595)	<i>F5</i> (Exon 13 (partial))	5 ml EDTA blood	2x/week	Sanger
Factor XII C46T (rs1801020)	<i>F12</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS

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Factor XIII V34L (rs5985)	<i>F13</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
β-Fibrinogen G-455A (rs1800790)	<i>FGB</i>	5 ml EDTA blood	2x/week	Sanger
FSAP G534E (rs7080536)	<i>HABP2</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
MTHFR C677T (rs1801133)	<i>MTHFR</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
MTHFR A1298C (rs1801131)	<i>MTHFR</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
PAI-1 A-844G (rs2227631)	<i>SERPINE1</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS
PAI-1 4G/5G (rs587776796)	<i>SERPINE1</i>	5 ml EDTA blood	2x/week	Multiplex-PCR, MALDI-TOF-MS

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.