

Metabolic Disorders / Internistic Syndromes

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
Meulengracht- (Gilbert-) Syndrome	<i>UGT1A1</i> *28 Polymorphism	5 ml EDTA blood	as needed 2-3 days	PCR, melting curve analysis
Cystic Fibrosis (CF) ⁺	<i>CFTR</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA
Hereditary Pancreatitis (HP) ⁺	<i>CFTR</i> (CDS complete + MLPA) <i>PRSS1</i> (CDS complete) <i>SPINK1</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA
Hereditary Periodic Fever Syndromes (HPFS) ⁺	<i>ELANE</i> (CDS complete) <i>IL1RN</i> (CDS complete) <i>IL36RN</i> (CDS complete) <i>LPIN2</i> (CDS complete) <i>MEFV</i> (CDS complete) <i>MVK</i> (CDS complete) <i>NLRC4</i> (CDS complete) <i>NLRP12</i> (CDS complete) <i>NLRP3</i> (CDS complete) <i>NOD2</i> (CDS complete) <i>PSMB8</i> (CDS complete) <i>PSTPIP1</i> (CDS complete) <i>TMEM173</i> (CDS complete) <i>TNFRSF1A</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Morbus Fabry ⁺	<i>GLA</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS

Metabolic Disorders / Internistic Syndromes

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)⁺	<i>ACADVL</i> (CDS complete) <i>ETFA</i> (CDS complete) <i>ETFB</i> (CDS complete) <i>ETFDH</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 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.