

Neurological Diseases

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
Hereditary Motor and Sensory Neuropathies (HMSN) ⁺, Charcot-Marie-Tooth Disease (CMT) ⁺	<i>FIG4</i> (CDS complete) <i>GDAP1</i> (CDS complete) <i>GJB1</i> (CDS complete) <i>IGHMBP2</i> (CDS complete) <i>LITAF</i> (CDS complete) <i>MFN2</i> (CDS complete) <i>MPZ</i> (CDS complete) <i>NEFL</i> (CDS complete) <i>PMP22</i> (CDS complete + MLPA) <i>SH3TC2</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA
Hereditary Spastic Paraplegia (HSP) ⁺, Strümpell-Lorrain Disease ⁺	<i>ABCD1</i> (CDS complete) <i>ATL1</i> (CDS complete) <i>CYP27A1</i> (CDS complete) <i>FA2H</i> (CDS complete) <i>KIF5A</i> (CDS complete) <i>PLP1</i> (CDS complete) <i>REEP1</i> (CDS complete) <i>SPAST</i> (CDS complete) <i>SPG7</i> (CDS complete) <i>SPG11</i> (CDS complete) <i>TTR</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Juvenile Parkinson's Disease ⁺	<i>ATP13A2</i> (CDS komplett) <i>PARK7</i> (CDS komplett) <i>PINK1</i> (CDS komplett) <i>PRKN</i> (CDS komplett)	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.