

**Muscle Diseases**

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
<b>Bethlem Myopathy</b> <sup>+</sup>	<i>COL6A1</i> (CDS complete) <i>COL6A2</i> (CDS complete) <i>COL6A3</i> (CDS complete) <i>CRYAB</i> (CDS complete) <i>PYGM</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
<b>Limb-Girdle Muscular Dystrophy (LGMD)</b> <sup>+</sup>	<i>ANO5</i> (CDS complete) <i>CAPN3</i> (CDS complete) <i>DYSF</i> (CDS complete) <i>FKRP</i> (CDS complete) <i>SGCA</i> (CDS complete) <i>SGCB</i> (CDS complete) <i>SGCD</i> (CDS complete) <i>SGCG</i> (CDS complete) <i>SMCHD1</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
<b>Metabolic Myopathy</b> <sup>+</sup>	<i>ACADVL</i> (CDS complete) <i>CPT1A</i> (CDS complete) <i>CPT2</i> (CDS complete) <i>ETFA</i> (CDS complete) <i>ETFB</i> (CDS complete) <i>ETFDH</i> (CDS complete) <i>GAA</i> (CDS complete) <i>LPIN1</i> (CDS complete) <i>PYGM</i> (CDS complete) <i>SLC22A5</i> (CDS complete) <i>SLC25A20</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS

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<b>Neutral Lipid Storage Disease with Myopathy (NLSDM) <sup>+</sup>, Chanarin-Dorfman Syndrome <sup>+</sup></b>	<i>ABHD5</i> (CDS complete) <i>ACADM</i> (CDS complete) <i>ACADS</i> (CDS complete) <i>ACADVL</i> (CDS complete) <i>CPT2</i> (CDS complete) <i>ETFA</i> (CDS complete) <i>ETFB</i> (CDS complete) <i>ETFDH</i> (CDS complete) <i>HADH</i> (CDS complete) <i>HADHA</i> (CDS complete) <i>HADHB</i> (CDS complete) <i>PNPLA2</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.