

GENETIC TEST: Myopathy (gene panel)

FULL NAME:	Myopathy (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region Whole Exome Sequencing (WES)
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
EQA:	• Muscular dystrophies
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	11 Dec 2020 - 09:55
CHANGED:	22 Jun 2022 - 11:04

URL:	https://labogidsmedgen.uza.be/analyses/whole-exome-sequencing-wes-ikv-myopathie...
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Related Analytes

- [abhydrolase domain containing 5, lysophosphatidic acid acyltransferase](#)
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- [acyl-CoA dehydrogenase short chain](#)
- [acyl-CoA dehydrogenase very long chain](#)
- [actin alpha 1, skeletal muscle](#)
- [actinin alpha 2](#)
- [adenylosuccinate synthase 1](#)
- [acylglycerol kinase](#)
- [amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase](#)
- [agrin](#)
- [aldolase, fructose-bisphosphate A](#)
- [ALG14 UDP-N-acetylglucosaminyltransferase subunit](#)
- [ALG2 alpha-1,3/1,6-mannosyltransferase](#)
- [anoctamin 5](#)
- [annexin A11](#)
- [activating signal cointegrator 1 complex subunit 1](#)
- [ATPase Na⁺/K⁺ transporting subunit alpha 2](#)
- [ATPase sarcoplasmic/endoplasmic reticulum Ca²⁺ transporting 1](#)
- [beta-1,3-N-acetylgalactosaminyltransferase 2](#)

- beta-1,4-glucuronyltransferase 1
- BAG cochaperone 3
- bridging integrator 1
- blood vessel epicardial substance
- calcium voltage-gated channel subunit alpha1 S
- cyclase associated actin cytoskeleton regulatory protein 2
- calpain 3
- calsequestrin 1
- caveolin 3
- caveolae associated protein 1
- cofilin 2
- choline O-acetyltransferase
- chromodomain helicase DNA binding protein 7
- choline kinase beta
- cholinergic receptor nicotinic alpha 1 subunit
- cholinergic receptor nicotinic beta 1 subunit
- cholinergic receptor nicotinic delta subunit
- cholinergic receptor nicotinic epsilon subunit
- cholinergic receptor nicotinic gamma subunit
- chloride voltage-gated channel 1
- CCHC-type zinc finger nucleic acid binding protein
- collagen type XII alpha 1 chain
- collagen type XIII alpha 1 chain
- collagen type XXV alpha 1 chain
- collagen type VI alpha 1 chain
- collagen type VI alpha 2 chain
- collagen type VI alpha 3 chain
- collagen like tail subunit of asymmetric acetylcholinesterase
- cytochrome c oxidase subunit 6A2
- carnitine palmitoyltransferase 2
- CDP-L-ribitol pyrophosphorylase A
- crystallin alpha B
- dystroglycan 1

- DC-STAMP domain containing 2
- desmin
- deoxyguanosine kinase
- dystrophin
- DM1 protein kinase
- DnaJ heat shock protein family (Hsp40) member B6
- dynammin 2
- DNA methyltransferase 3 beta
- docking protein 7
- dolichyl-phosphate N-acetylglucosaminophosphotransferase 1
- dolichyl-phosphate mannosyltransferase subunit 2, regulatory
- dolichyl-phosphate mannosyltransferase subunit 3, regulatory
- dysferlin
- emerin
- enolase 3
- electron transfer flavoprotein subunit alpha
- electron transfer flavoprotein dehydrogenase
- FAM111 trypsin like peptidase B
- ferredoxin 2
- four and a half LIM domains 1
- fukutin related protein
- fukutin
- flavin adenine dinucleotide synthetase 1
- filamin C
- FMR1 autosomal homolog 1
- alpha glucosidase
- 1,4-alpha-glucan branching enzyme 1
- growth factor, augmenter of liver regeneration
- glutamine--fructose-6-phosphate transaminase 1
- geranylgeranyl diphosphate synthase 1
- GDP-mannose pyrophosphorylase B
- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase
- glycogenin 1

- glycogen synthase 1
- 3-hydroxyacyl-CoA dehydratase 1
- hydroxyacyl-CoA dehydrogenase
- hydroxyacyl-CoA dehydrogenase trifunctional multienzyme complex subunit alpha
- hydroxyacyl-CoA dehydrogenase trifunctional multienzyme complex subunit beta
- heterogeneous nuclear ribonucleoprotein A1
- heterogeneous nuclear ribonucleoprotein A2/B1
- heterogeneous nuclear ribonucleoprotein D like
- inositol polyphosphate-5-phosphatase K
- iron-sulfur cluster assembly enzyme
- integrin subunit alpha 7
- integrin subunit alpha 9
- jagged canonical Notch ligand 2
- kelch repeat and BTB domain containing 13
- potassium inwardly rectifying channel subfamily J member 2
- kinesin family member 21A
- kelch like family member 40
- kelch like family member 41
- kelch like family member 9
- kyphoscoliosis peptidase
- laminin subunit alpha 2
- laminin subunit beta 2
- lysosomal associated membrane protein 2
- LARGE xylosyl- and glucuronyltransferase 1
- LIM domain binding 3
- lactate dehydrogenase A
- LIM zinc finger domain containing 2
- lamin A/C
- leiomodlin 3
- lipin 1
- LDL receptor related protein 4
- mitogen-activated protein kinase kinase kinase 20
- matrin 3

- myoglobin
- multiple EGF like domains 10
- mitochondrial calcium uptake 1
- muscular LMNA interacting protein
- misato mitochondrial distribution and morphology regulator 1
- myotubularin 1
- muscle associated receptor tyrosine kinase
- myosin binding protein C1
- myosin binding protein C3
- myosin heavy chain 14
- myosin heavy chain 2
- myosin heavy chain 3
- myosin heavy chain 7
- myosin light chain 1
- myosin light chain 2
- myomixer, myoblast fusion factor
- myosin XVIIIIB
- myosin IXA
- myogenic differentiation 1
- myotilin
- myopalladin
- nebulin
- N-acetylneuraminate pyruvate lyase
- OPA1 mitochondrial dynamin like GTPase
- ORAI calcium release-activated calcium modulator 1
- poly(A) binding protein nuclear 1
- paired box 7
- phosphofructokinase, muscle
- phosphoglycerate mutase 2
- phosphoglycerate kinase 1
- phosphoglucomutase 1
- phosphorylase kinase regulatory subunit alpha 1
- phosphorylase kinase regulatory subunit beta

- paired like homeobox 2A
- plectin
- patatin like phospholipase domain containing 2
- patatin like phospholipase domain containing 8
- protein O-glucosyltransferase 1
- DNA polymerase gamma, catalytic subunit
- DNA polymerase gamma 2, accessory subunit
- protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-)
- protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1,4-)
- protein O-mannose kinase
- protein O-mannosyltransferase 1
- protein O-mannosyltransferase 2
- popeye domain containing 3
- prolyl endopeptidase like
- protein kinase AMP-activated non-catalytic subunit gamma 2
- glycogen phosphorylase, muscle associated
- pyridine nucleotide-disulphide oxidoreductase domain 1
- receptor associated protein of the synapse
- RANBP2-type and C3HC4-type zinc finger containing 1
- ribitol xylosyltransferase 1
- ryanodine receptor 1
- sodium voltage-gated channel alpha subunit 4
- selenoprotein N
- sarcoglycan alpha
- sarcoglycan beta
- sarcoglycan delta
- sarcoglycan gamma
- SIL1 nucleotide exchange factor
- solute carrier family 18 member A3
- solute carrier family 22 member 5
- solute carrier family 25 member 20
- solute carrier family 25 member 4
- solute carrier family 5 member 7

- structural maintenance of chromosomes flexible hinge domain containing 1
- small muscle protein X-linked
- striated muscle enriched protein kinase
- sequestosome 1
- SH3 and cysteine rich domain 3
- stromal interaction molecule 1
- supervillin
- spectrin repeat containing nuclear envelope protein 1
- spectrin repeat containing nuclear envelope protein 2
- synaptotagmin 2
- titin-cap
- TIA1 cytotoxic granule associated RNA binding protein
- thymidine kinase 2
- transmembrane protein 126B
- transmembrane protein 43
- troponin C2, fast skeletal type
- troponin I2, fast skeletal type
- troponin T1, slow skeletal type
- troponin T3, fast skeletal type
- transportin 3
- tenascin XB
- DNA topoisomerase III alpha
- torsin 1A interacting protein 1
- triosephosphate isomerase 1
- tropomyosin 2
- tropomyosin 3
- trafficking protein particle complex subunit 11
- triadin
- tripartite motif containing 32
- thyroid hormone receptor interactor 4
- tRNA methyltransferase 5
- titin
- tubulin beta 3 class III

- twinkle mtDNA helicase
- unc-45 myosin chaperone B
- vesicle associated membrane protein 1
- valosin containing protein
- vacuolar ATPase assembly factor VMA21
- von Willebrand factor A domain containing 1

Related Gene Panels

- Myopathy (genepanel) - UZA

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