

GENETIC TEST: Neuromuscular disorders (gene panel)

FULL NAME:	Neuromuscular disorders (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) Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-09-11 / 2026-09-10
EQA:	• Muscular dystrophies
TURNAROUND TIME (MAXIMUM):	6 months

CREATED:	31 Jul 2019 - 14:35
CHANGED:	16 Dec 2022 - 12:43

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RELATED CONTENT

Related Laboratories

- [Centrum Medische Genetica - UZ Gent](#)

Related Analytes

- [abhydrolase domain containing 5, lysophosphatidic acid acyltransferase](#)
- [acyl-CoA dehydrogenase family member 9](#)
- [acyl-CoA dehydrogenase long chain](#)
- [acyl-CoA dehydrogenase medium chain](#)
- [acyl-CoA dehydrogenase short chain](#)
- [acyl-CoA dehydrogenase very long chain](#)
- [actin alpha 1, skeletal muscle](#)
- [actinin alpha 2](#)
- [activin A receptor type 1](#)
- [adenylate cyclase 6](#)
- [adhesion G protein-coupled receptor G6](#)
- [adenylosuccinate synthase 1](#)
- [amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase](#)
- [agrin](#)
- [aldolase, fructose-bisphosphate A](#)
- [ALG13 UDP-N-acetylglucosaminyltransferase subunit](#)
- [ALG14 UDP-N-acetylglucosaminyltransferase subunit](#)
- [ALG2 alpha-1,3/1,6-mannosyltransferase](#)
- [adenosine monophosphate deaminase 1](#)
- [anoctamin 5](#)

- apolipoprotein O
- N-acylsphingosine amidohydrolase 1
- activating signal cointegrator 1 complex subunit 1
- aspartate beta-hydroxylase
- ATPase sarcoplasmic/endoplasmic reticulum Ca²⁺ transporting 1
- beta-1,3-N-acetylgalactosaminyltransferase 2
- beta-1,4-glucuronyltransferase 1
- BAG cochaperone 3
- Bet1 golgi vesicular membrane trafficking protein
- bridging integrator 1
- BSCL2 lipid droplet biogenesis associated, seipin
- blood vessel epicardial substance
- calcium voltage-gated channel subunit alpha1 H
- calcium voltage-gated channel subunit alpha1 S
- calpain 3
- calsequestrin 1
- caveolin 3
- caveolae associated protein 1
- coiled-coil domain containing 78
- complement factor H
- cofilin 2
- choline O-acetyltransferase
- coiled-coil-helix-coiled-coil-helix domain containing 10
- 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
- CLN3 lysosomal/endosomal transmembrane protein, battenin
- caseinolytic mitochondrial matrix peptidase proteolytic subunit
- contactin 1

- collagen type XII alpha 1 chain
- collagen type XIII alpha 1 chain
- collagen type IV alpha 1 chain
- collagen type VI alpha 1 chain
- collagen type VI alpha 2 chain
- collagen type VI alpha 3 chain
- collagen type IX alpha 3 chain
- collagen like tail subunit of asymmetric acetylcholinesterase
- corin, serine peptidase
- carnitine palmitoyltransferase 2
- CDP-L-ribitol pyrophosphorylase A
- crystallin alpha B
- dystroglycan 1
- desmin
- deoxyguanosine kinase
- DEAH-box helicase 16
- dystrophin
- DM1 protein kinase
- DnaJ heat shock protein family (Hsp40) member B6
- dynammin 2
- DNA methyltransferase 3 beta
- docking protein 7
- dolichol kinase
- dolichyl-phosphate N-acetylglucosaminophosphotransferase 1
- dolichyl-phosphate mannosyltransferase subunit 1, catalytic
- dolichyl-phosphate mannosyltransferase subunit 2, regulatory
- dolichyl-phosphate mannosyltransferase subunit 3, regulatory
- dysferlin
- endothelin converting enzyme like 1
- emerin
- enolase 3
- ectopic P-granules 5 autophagy tethering factor
- electron transfer flavoprotein subunit alpha

- electron transfer flavoprotein subunit beta
- electron transfer flavoprotein dehydrogenase
- FAM111 trypsin like peptidase B
- ferredoxin 2
- FKBP prolyl isomerase 14
- 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
- gliomedin
- GLE1 RNA export mediator
- GDP-mannose pyrophosphorylase B
- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase
- golgin A2
- golgi SNAP receptor complex member 2
- glycogenin 1
- glycogen synthase 1
- 3-hydroxyacyl-CoA dehydratase 1
- hydroxyacyl-CoA dehydrogenase trifunctional multienzyme complex subunit alpha
- hydroxyacyl-CoA dehydrogenase trifunctional multienzyme complex subunit beta
- hexosaminidase subunit beta
- heterogeneous nuclear ribonucleoprotein A1
- heterogeneous nuclear ribonucleoprotein A2/B1
- heterogeneous nuclear ribonucleoprotein D like
- HRas proto-oncogene, GTPase
- heat shock protein family B (small) member 1
- heat shock protein family B (small) member 3

- heat shock protein family B (small) member 8
- heparan sulfate proteoglycan 2
- HtrA serine peptidase 2
- inositol polyphosphate-5-phosphatase K
- iron-sulfur cluster assembly enzyme
- integrin subunit alpha 7
- kelch repeat and BTB domain containing 13
- kelch like family member 40
- kelch like family member 41
- kelch like family member 9
- kyphoscoliosis peptidase
- laminin subunit alpha 2
- laminin subunit alpha 5
- laminin subunit beta 2
- lysosomal associated membrane protein 2
- LARGE xylosyl- and glucuronyltransferase 1
- leucyl-tRNA synthetase 2, mitochondrial
- LAS1 like ribosome biogenesis factor
- LIM domain binding 3
- lactate dehydrogenase A
- leucine rich repeat LGI family member 4
- LIM zinc finger domain containing 2
- lamin A/C
- leiomodin 3
- lysyl oxidase like 4
- lipin 1
- ligand dependent nuclear receptor interacting factor 1
- mitogen-activated protein kinase kinase kinase 20
- myoglobin
- mucolipin TRP cation channel 1
- multiple EGF like domains 10
- MET proto-oncogene, receptor tyrosine kinase
- mitochondrial calcium uptake 1

- mannose-P-dolichol utilization defect 1
- mitochondrial ribosomal protein S25
- myostatin
- misato mitochondrial distribution and morphology regulator 1
- myotubularin 1
- myotubularin related protein 14
- muscle associated receptor tyrosine kinase
- myosin binding protein C1
- myosin binding protein C3
- myogenic factor 5
- myosin heavy chain 2
- myosin heavy chain 3
- myosin heavy chain 7
- myosin heavy chain 7B
- myosin heavy chain 8
- myosin light chain 1
- myosin light chain 2
- myomaker, myoblast fusion factor
- myosin XVIII B
- myosin IXA
- myotilin
- myopalladin
- nebulin
- neurofilament light chain
- ORAI calcium release-activated calcium modulator 1
- poly(A) binding protein nuclear 1
- paired box 7
- peroxisomal biogenesis factor 6
- phosphofructokinase, muscle
- phosphoglycerate mutase 2
- phosphoglycerate kinase 1
- phosphoglucomutase 1
- phosphorylase kinase regulatory subunit alpha 1

- phosphorylase kinase regulatory subunit beta
- piezo type mechanosensitive ion channel component 2
- 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
- peptidyl-tRNA hydrolase 2
- pseudouridine synthase 1
- 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
- ribonucleotide reductase regulatory TP53 inducible subunit M2B
- ribitol xylosyltransferase 1
- ryanodine receptor 1
- ryanodine receptor 3
- sodium voltage-gated channel alpha subunit 4
- selenoprotein N
- sarcoglycan alpha
- sarcoglycan beta
- sarcoglycan delta
- sarcoglycan gamma
- SIL1 nucleotide exchange factor

- solute carrier family 16 member 1
- solute carrier family 18 member A3
- solute carrier family 22 member 5
- solute carrier family 25 member 1
- solute carrier family 25 member 20
- solute carrier family 25 member 4
- solute carrier family 25 member 42
- solute carrier family 5 member 7
- structural maintenance of chromosomes flexible hinge domain containing 1
- small muscle protein X-linked
- synaptosome associated protein 25
- striated muscle enriched protein kinase
- spectrin beta, non-erythrocytic 4
- sequestosome 1
- SH3 and cysteine rich domain 3
- stromal interaction molecule 1
- succinate-CoA ligase ADP-forming subunit beta
- supervillin
- spectrin repeat containing nuclear envelope protein 1
- spectrin repeat containing nuclear envelope protein 2
- synaptotagmin 2
- TAR DNA binding protein
- titin-cap
- transcription factor A, mitochondrial
- TIA1 cytotoxic granule associated RNA binding protein
- translocase of inner mitochondrial membrane 22
- thymidine kinase 2
- transmembrane protein 43
- transmembrane protein 65
- troponin C2, fast skeletal type
- troponin I2, fast skeletal type
- troponin T1, slow skeletal type
- troponin T3, fast skeletal type

- [transportin 3](#)
- [tenascin XB](#)
- [torsin 1A interacting protein 1](#)
- [tropomyosin 2](#)
- [tropomyosin 3](#)
- [trafficking protein particle complex subunit 11](#)
- [tripartite motif containing 32](#)
- [tripartite motif containing 54](#)
- [thyroid hormone receptor interactor 4](#)
- [tRNA splicing endonuclease subunit 54](#)
- [Ts translation elongation factor, mitochondrial](#)
- [titin](#)
- [thymidine phosphorylase](#)
- [unc-45 myosin chaperone B](#)
- [vesicle associated membrane protein 1](#)
- [valosin containing protein](#)
- [vacuolar ATPase assembly factor VMA21](#)
- [VPS33B late endosome and lysosome associated](#)
- [tyrosyl-tRNA synthetase 2](#)
- [zinc finger and BTB domain containing 42](#)
- [zinc finger C4H2-type containing](#)

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