

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, DNA
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2022-10-07 / 2023-10-06
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	08 Aug 2019 - 08:55
CHANGED:	24 Jan 2023 - 11:53

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RELATED CONTENT

Related Diseases

- [Becker muscular dystrophy](#)
- [Duchenne muscular dystrophy](#)

Related Laboratories

- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)

Related Analytes

- [alanyl-tRNA synthetase 1](#)
- [abhydrolase domain containing 5, lysophosphatidic acid acyltransferase](#)
- [acyl-CoA dehydrogenase family member 9](#)
- [acyl-CoA dehydrogenase medium chain](#)
- [acyl-CoA dehydrogenase very long chain](#)
- [actin alpha 1, skeletal muscle](#)
- [activin A receptor type 1](#)
- [adenylosuccinate synthase 1](#)
- [acylglycerol kinase](#)
- [amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase](#)
- [agrin](#)
- [apoptosis inducing factor mitochondria associated 1](#)
- [aldolase, fructose-bisphosphate A](#)
- [ALG13 UDP-N-acetylglucosaminyltransferase subunit](#)

- ALG14 UDP-N-acetylglucosaminyltransferase subunit
- ALG2 alpha-1,3/1,6-mannosyltransferase
- alsin Rho guanine nucleotide exchange factor ALS2
- adenosine monophosphate deaminase 1
- angiogenin
- anoctamin 5
- androgen receptor
- N-acylsphingosine amidohydrolase 1
- activating signal cointegrator 1 complex subunit 1
- ATPase sarcoplasmic/endoplasmic reticulum Ca2+ transporting 1
- ATPase copper transporting alpha
- ataxin 2
- beta-1,3-N-acetylgalactosaminyltransferase 2
- beta-1,4-glucuronyltransferase 1
- BAG cochaperone 3
- BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone
- BICD cargo adaptor 2
- bridging integrator 1
- BSCL2 lipid droplet biogenesis associated, seipin
- blood vessel epicardial substance
- complement C1q binding protein
- C9orf72-SMCR8 complex subunit
- calcium voltage-gated channel subunit alpha1 A
- calcium voltage-gated channel subunit alpha1 S
- calpain 3
- calsequestrin 1
- caveolin 3
- caveolae associated protein 1
- coiled-coil domain containing 78
- cofilin 2
- choline O-acetyltransferase
- coiled-coil-helix-coiled-coil-helix domain containing 10
- choline kinase beta

- charged multivesicular body protein 2B
- 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
- carbohydrate sulfotransferase 14
- chloride voltage-gated channel 1
- CLN3 lysosomal/endosomal transmembrane protein, battenin
- CCHC-type zinc finger nucleic acid binding protein
- contactin 1
- collagen type XII alpha 1 chain
- collagen type XIII 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
- coenzyme Q9
- cytochrome c oxidase assembly factor heme A:farnesyltransferase COX10
- cytochrome c oxidase assembly homolog COX15
- cytochrome c oxidase assembly factor COX20
- cytochrome c oxidase subunit 6B1
- cytochrome c oxidase subunit 8A
- carnitine palmitoyltransferase 2
- CDP-L-ribitol pyrophosphorylase A
- crystallin alpha B
- dystroglycan 1
- dynactin subunit 1
- desmin
- deoxyguanosine kinase
- dystrophin
- DM1 protein kinase
- DNA replication helicase/nuclease 2

- DnaJ heat shock protein family (Hsp40) member B2
- 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 1, catalytic
- dolichyl-phosphate mannosyltransferase subunit 2, regulatory
- dolichyl-phosphate mannosyltransferase subunit 3, regulatory
- dynein cytoplasmic 1 heavy chain 1
- dysferlin
- endothelin converting enzyme like 1
- emerin
- enolase 3
- erb-b2 receptor tyrosine kinase 3
- erb-b2 receptor tyrosine kinase 4
- electron transfer flavoprotein subunit alpha
- electron transfer flavoprotein subunit beta
- electron transfer flavoprotein dehydrogenase
- exosome component 3
- exosome component 8
- FAST kinase domains 2
- fibrillin 2
- F-box and leucine rich repeat protein 4
- F-box protein 38
- ferredoxin 2
- four and a half LIM domains 1
- FIG4 phosphoinositide 5-phosphatase
- fukutin related protein
- fukutin
- flavin adenine dinucleotide synthetase 1
- filamin C
- FAD dependent oxidoreductase domain containing 1

- FUS RNA binding protein
- FMR1 autosomal homolog 1
- alpha glucosidase
- gamma-aminobutyric acid type A receptor subunit alpha3
- glycyl-tRNA synthetase 1
- 1,4-alpha-glucan branching enzyme 1
- growth factor, augments liver regeneration
- glutamine--fructose-6-phosphate transaminase 1
- GLE1 RNA export mediator
- GLI family zinc finger 3
- GDP-mannose pyrophosphorylase B
- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase
- golgin A2
- 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 3
- heat shock protein family B (small) member 8
- heparan sulfate proteoglycan 2
- iron-sulfur cluster assembly factor IBA57
- immunoglobulin mu DNA binding protein 2
- inositol polyphosphate-5-phosphatase K
- iron-sulfur cluster assembly enzyme
- integrin subunit alpha 7
- kelch repeat and BTB domain containing 13
- potassium voltage-gated channel subfamily A member 1

- potassium voltage-gated channel subfamily H member 2
- potassium inwardly rectifying channel subfamily J member 18
- potassium inwardly rectifying channel subfamily J member 2
- potassium voltage-gated channel subfamily Q member 1
- kinesin family member 5A
- 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
- 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
- lipin 1
- LDL receptor related protein 4
- leucine rich pentatricopeptide repeat containing
- mitogen-activated protein kinase kinase kinase 20
- matrin 3
- methylcrotonyl-CoA carboxylase subunit 1
- methylcrotonyl-CoA carboxylase subunit 2
- multiple EGF like domains 10
- mitochondrial genome maintenance exonuclease 1
- mitochondrial inner membrane protein MPV17
- myostatin
- myotubularin 1
- myotubularin related protein 14

- muscle associated receptor tyrosine kinase
- myosin binding protein C1
- myosin binding protein C3
- myosin heavy chain 2
- myosin heavy chain 3
- myosin heavy chain 7
- myosin heavy chain 8
- myosin light chain 1
- myomaker, myoblast fusion factor
- myosin XVIIIIB
- myosin IXA
- myotilin
- myopalladin
- NADH:ubiquinone oxidoreductase subunit A1
- NADH:ubiquinone oxidoreductase subunit A10
- NADH:ubiquinone oxidoreductase subunit A11
- NADH:ubiquinone oxidoreductase subunit A12
- NADH:ubiquinone oxidoreductase subunit A2
- NADH:ubiquinone oxidoreductase subunit A3
- NADH:ubiquinone oxidoreductase subunit A9
- NADH:ubiquinone oxidoreductase complex assembly factor 1
- NADH:ubiquinone oxidoreductase complex assembly factor 2
- NADH:ubiquinone oxidoreductase complex assembly factor 5
- NADH:ubiquinone oxidoreductase complex assembly factor 6
- NADH:ubiquinone oxidoreductase core subunit S1
- NADH:ubiquinone oxidoreductase core subunit S2
- NADH:ubiquinone oxidoreductase core subunit S3
- NADH:ubiquinone oxidoreductase subunit S4
- NADH:ubiquinone oxidoreductase core subunit S7
- NADH:ubiquinone oxidoreductase core subunit S8
- NADH:ubiquinone oxidoreductase core subunit V1
- NADH:ubiquinone oxidoreductase core subunit V2
- nebulin

- neurofilament heavy chain
- NUBP iron-sulfur cluster assembly factor, mitochondrial
- optineurin
- ORAI calcium release-activated calcium modulator 1
- poly(A) binding protein nuclear 1
- PET100 cytochrome c oxidase chaperone
- phosphofructokinase, muscle
- profilin 1
- phosphoglycerate mutase 2
- phosphoglycerate kinase 1
- phosphoglucomutase 1
- phosphorylase kinase regulatory subunit alpha 1
- phosphorylase kinase regulatory subunit beta
- phosphorylase kinase catalytic subunit gamma 1
- piezo type mechanosensitive ion channel component 2
- phosphatidylinositol-4-phosphate 5-kinase type 1 gamma
- plectin
- pleckstrin homology and RhoGEF domain containing G5
- 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
- prolyl endopeptidase like
- protein kinase AMP-activated non-catalytic subunit gamma 2
- peripherin
- 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
- RNA binding motif protein 7
- receptor accessory protein 1
- ribonuclease H1
- ribonucleotide reductase regulatory TP53 inducible subunit M2B
- ribitol xylosyltransferase 1
- ryanodine receptor 1
- sodium voltage-gated channel alpha subunit 4
- synthesis of cytochrome C oxidase 1
- synthesis of cytochrome C oxidase 2
- selenoprotein N
- senataxin
- sarcoglycan alpha
- sarcoglycan beta
- sarcoglycan delta
- sarcoglycan gamma
- sigma non-opioid intracellular receptor 1
- 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 52 member 2
- solute carrier family 52 member 3
- solute carrier family 5 member 7
- structural maintenance of chromosomes flexible hinge domain containing 1
- survival of motor neuron 1, telomeric
- synaptosome associated protein 25
- superoxide dismutase 1
- striated muscle enriched protein kinase
- SPG11 vesicle trafficking associated, spatacsin

- SPG7 matrix AAA peptidase subunit, paraplegin
- spectrin beta, non-erythrocytic 4
- sequestosome 1
- SH3 and cysteine rich domain 3
- stromal interaction molecule 1
- succinate-CoA ligase ADP-forming subunit beta
- succinate-CoA ligase GDP/ADP-forming subunit alpha
- SURF1 cytochrome c oxidase assembly factor
- spectrin repeat containing nuclear envelope protein 1
- spectrin repeat containing nuclear envelope protein 2
- synaptotagmin 2
- translational activator of cytochrome c oxidase I
- tafazzin, phospholipid-lysophospholipid transacylase
- transport and golgi organization 2 homolog
- TAR DNA binding protein
- threonyl-tRNA synthetase 2, mitochondrial
- titin-cap
- TIA1 cytotoxic granule associated RNA binding protein
- thymidine kinase 2
- transmembrane protein 43
- troponin I2, fast skeletal type
- troponin T1, slow skeletal type
- troponin T3, fast skeletal type
- transportin 3
- DNA topoisomerase III alpha
- torsin 1A interacting protein 1
- tropomyosin 2
- tropomyosin 3
- trafficking protein particle complex subunit 11
- tripartite motif containing 32
- tripartite motif containing 54
- tripartite motif containing 63
- thyroid hormone receptor interactor 4

- [tRNA methyltransferase 5](#)
- [transient receptor potential cation channel subfamily V member 4](#)
- [Ts translation elongation factor, mitochondrial](#)
- [titin](#)
- [tubulin alpha 4a](#)
- [twinkle mtDNA helicase](#)
- [thymidine phosphorylase](#)
- [ubiquitin like modifier activating enzyme 1](#)
- [ubiquilin 2](#)
- [vesicle associated membrane protein 1](#)
- [VAMP associated protein B and C](#)
- [valyl-tRNA synthetase 1](#)
- [valosin containing protein](#)
- [vacuolar ATPase assembly factor VMA21](#)
- [VPS33B late endosome and lysosome associated](#)
- [VRK serine/threonine kinase 1](#)
- [tryptophanyl-tRNA synthetase 1](#)
- [X-linked Kx blood group antigen, Kell and VPS13A binding protein](#)
- [tyrosyl-tRNA synthetase 2](#)
- [zinc finger C4H2-type containing](#)

Related Gene Panels

- [Myopathy \(332 genes\) - IPG](#)

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