

GENETIC TEST:

Neuromuscular disorders (548 genes)

FULL NAME:	Neuromuscular disorders (548 genes)
DESCRIPTION:	Muscular dystrophy (Congenital), Myopathy (congenital, distal, others), Myotonic syndrome, Muscle channelopathies, Metabolic myopathy, Congenital myasthenic syndrome, Motor neuron diseases, Hereditary motor and sensory neuropathy, Malignant hyperthermia, other neuromuscular disorder
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 Mendeliome
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-03-25 / 2024-09-09

EQA:	• Muscular dystrophies
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	22 Aug 2019 - 14:42
CHANGED:	23 Jan 2023 - 15:13
URL:	http://erasmeinfo.ulb.ac.be/Compendium/scripts/biolist.idc?Parlistanal=201908

Source URL: http://gentest.healthdata.be/genetic_test/619

RELATED CONTENT

Related Laboratories

- [Centre de Génétique Humaine - Erasme ULB](#)

Related Analytes

- [alanyl-tRNA synthetase 1](#)
- [ATP binding cassette subfamily D member 1](#)
- [abhydrolase domain containing 12, lysophospholipase](#)
- [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](#)
- [activin A receptor type 1](#)
- [adenosine deaminase RNA specific](#)
- [adenylosuccinate synthase 1](#)
- [AFG3 like matrix AAA peptidase subunit 2](#)
- [acylglycerol kinase](#)
- [amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase](#)
- [agrin](#)
- [apoptosis inducing factor mitochondria associated 1](#)
- [aminoacyl tRNA synthetase complex interacting multifunctional protein 1](#)
- [aldehyde dehydrogenase 18 family member A1](#)

- aldehyde dehydrogenase 3 family member A2
- 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
- adenosine monophosphate deaminase 2
- angiogenin
- anoctamin 5
- annexin A11
- adaptor related protein complex 4 subunit beta 1
- adaptor related protein complex 4 subunit epsilon 1
- adaptor related protein complex 4 subunit mu 1
- adaptor related protein complex 4 subunit sigma 1
- adaptor related protein complex 5 subunit zeta 1
- aprataxin
- androgen receptor
- arginase 1
- Rho guanine nucleotide exchange factor 10
- Rho guanine nucleotide exchange factor 28
- ADP ribosylation factor like GTPase 6 interacting protein 1
- arylsulfatase family member I
- N-acylsphingosine amidohydrolase 1
- activating signal cointegrator 1 complex subunit 1
- atlastin GTPase 1
- atlastin GTPase 3
- ATM serine/threonine kinase
- ATPase cation transporting 13A2
- ATPase Na⁺/K⁺ transporting subunit alpha 1
- ATPase sarcoplasmic/endoplasmic reticulum Ca²⁺ transporting 1
- ATPase copper transporting alpha
- ataxin 2

- AU RNA binding methylglutaconyl-CoA hydratase
- beta-1,3-N-acetylgalactosaminyltransferase 2
- beta-1,4-N-acetyl-galactosaminyltransferase 1
- 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
- chromosome 19 open reading frame 12
- complement C1q binding protein
- C9orf72-SMCR8 complex subunit
- calcium voltage-gated channel subunit alpha1 A
- calcium voltage-gated channel subunit alpha1 S
- calpain 1
- calpain 3
- calsequestrin 1
- caveolin 3
- caveolae associated protein 1
- coiled-coil domain containing 78
- chaperonin containing TCP1 subunit 5
- complement factor H
- 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
- CLN8 transmembrane ER and ERGIC protein
- clathrin heavy chain like 1
- CCHC-type zinc finger nucleic acid binding protein
- contactin 1
- contactin associated protein 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
- corin, serine peptidase
- 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 6A1
- cytochrome c oxidase subunit 6B1
- cytochrome c oxidase subunit 8A
- carnitine palmitoyltransferase 1C
- carnitine palmitoyltransferase 2
- CDP-L-ribitol pyrophosphorylase A
- crystallin alpha B
- colony stimulating factor 1 receptor
- CTD phosphatase subunit 1
- cytochrome P450 family 27 subfamily A member 1
- cytochrome P450 family 2 subfamily U member 1
- cytochrome P450 family 7 subfamily B member 1
- dystroglycan 1
- aspartyl-tRNA synthetase 2, mitochondrial

- DDB1 and CUL4 associated factor 8
- dynactin subunit 1
- DDHD domain containing 1
- DDHD domain containing 2
- desmin
- diacylglycerol O-acyltransferase 2
- deoxyguanosine kinase
- dehydrogenase E1 and transketolase domain containing 1
- dystrophin
- DM1 protein kinase
- Dmx like 2
- DnaJ heat shock protein family (Hsp40) member B2
- DnaJ heat shock protein family (Hsp40) member B6
- dynamain 2
- DNA methyltransferase 1
- 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
- dystonin
- dynein cytoplasmic 1 heavy chain 1
- dysferlin
- endothelin converting enzyme like 1
- early growth response 2
- ELOVL fatty acid elongase 4
- elongator complex protein 1
- emerin
- enolase 3
- ectonucleoside triphosphate diphosphohydrolase 1
- erb-b2 receptor tyrosine kinase 3
- erb-b2 receptor tyrosine kinase 4

- ERCC excision repair 2, TFIIH core complex helicase subunit
- ER lipid raft associated 1
- ER lipid raft associated 2
- electron transfer flavoprotein subunit alpha
- electron transfer flavoprotein subunit beta
- electron transfer flavoprotein dehydrogenase
- exosome component 3
- exosome component 8
- fatty acid 2-hydroxylase
- FAM111 trypsin like peptidase B
- phenylalanyl-tRNA synthetase 2, mitochondrial
- FAST kinase domains 2
- fibulin 5
- fibrillin 2
- F-box and leucine rich repeat protein 4
- F-box protein 38
- F-box protein 7
- ferredoxin 2
- FYVE, RhoGEF and PH domain containing 4
- FIG4 phosphoinositide 5-phosphatase
- fukutin related protein
- fukutin
- flavin adenine dinucleotide synthetase 1
- filamin C
- fibronectin leucine rich transmembrane protein 1
- FLVCR heme transporter 1
- FAD dependent oxidoreductase domain containing 1
- FUS RNA binding protein
- frataxin
- alpha glucosidase
- gamma-aminobutyric acid type A receptor subunit alpha3
- glutamate decarboxylase 1
- galactosylceramidase

- gigaxonin
- glycyl-tRNA synthetase 1
- phosphoribosylglycinamide formyltransferase, phosphoribosylglycinamide synthetase, phosphoribosylaminoimidazole synthetase
- glucosylceramidase beta 1
- glucosylceramidase beta 2
- 1,4-alpha-glucan branching enzyme 1
- GTP cyclohydrolase 1
- ganglioside induced differentiation associated protein 1
- glial fibrillary acidic protein
- growth factor, augmenter of liver regeneration
- glutamine--fructose-6-phosphate transaminase 1
- geranylgeranyl diphosphate synthase 1
- gap junction protein beta 1
- gap junction protein beta 3
- gap junction protein gamma 2
- galactosidase alpha
- galactosidase beta 1
- GLE1 RNA export mediator
- GLI family zinc finger 3
- glycolipid transfer protein
- GDP-mannose pyrophosphorylase B
- G protein subunit beta 4
- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase
- golgin A2
- golgi SNAP receptor complex member 2
- gelsolin
- 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
- histidyl-tRNA synthetase 1
- hepatic and glial cell adhesion molecule

- hexosaminidase subunit alpha
- hexosaminidase subunit beta
- histidine triad nucleotide binding protein 1
- hexokinase 1
- heterogeneous nuclear ribonucleoprotein A1
- heterogeneous nuclear ribonucleoprotein A2/B1
- heterogeneous nuclear ribonucleoprotein D like
- homeobox D10
- HRas proto-oncogene, GTPase
- hydroxysteroid 17-beta dehydrogenase 4
- heat shock protein family B (small) member 1
- heat shock protein family B (small) member 3
- heat shock protein family B (small) member 8
- heat shock protein family D (Hsp60) member 1
- heparan sulfate proteoglycan 2
- hyccin PI4KA lipid kinase complex subunit 1
- iron-sulfur cluster assembly factor IBA57
- interferon induced with helicase C domain 1
- interferon related developmental regulator 1
- immunoglobulin mu DNA binding protein 2
- inverted formin, FH2 and WH2 domain containing
- inositol polyphosphate-5-phosphatase K
- iron-sulfur cluster assembly enzyme
- integrin subunit alpha 7
- lysyl-tRNA synthetase 1
- kelch repeat and BTB domain containing 13
- potassium voltage-gated channel subfamily A member 1
- potassium voltage-gated channel subfamily E regulatory subunit 1
- potassium voltage-gated channel subfamily E regulatory subunit 2
- potassium voltage-gated channel subfamily E regulatory subunit 3
- potassium voltage-gated channel subfamily H member 2
- potassium inwardly rectifying channel subfamily J member 12
- potassium inwardly rectifying channel subfamily J member 2

- potassium voltage-gated channel subfamily Q member 1
- kinesin family member 1A
- kinesin family member 1B
- kinesin family member 1C
- kinesin family member 21A
- kinesin family member 5A
- kinesin light chain 4
- kelch like family member 40
- kelch like family member 41
- kelch like family member 9
- kyphoscoliosis peptidase
- L1 cell adhesion molecule
- 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
- lipopolysaccharide induced TNF factor
- lamin A/C
- leiomodlin 3
- lipin 1
- leucine rich pentatricopeptide repeat containing
- leucine rich repeat and sterile alpha motif containing 1
- mitogen-activated protein kinase kinase kinase 20
- methionyl-tRNA synthetase 1
- methionyl-tRNA synthetase 2, mitochondrial
- matrin 3
- myoglobin
- mediator complex subunit 12

- mediator complex subunit 25
- multiple EGF like domains 10
- mitofusin 2
- modulator of VRAC current 1
- membrane metalloendopeptidase
- MORC family CW-type zinc finger 2
- mitochondrial inner membrane protein MPV17
- myelin protein zero
- myostatin
- myotubularin 1
- myotubularin related protein 14
- myotubularin related protein 2
- mitochondrial poly(A) polymerase
- mitochondrial translation release factor in rescue
- muscle associated receptor tyrosine kinase
- myosin binding protein C1
- myosin binding protein C3
- myosin heavy chain 2
- myosin heavy chain 3
- myosin heavy chain 4
- myosin heavy chain 7
- myosin heavy chain 7B
- myosin heavy chain 8
- myosin light chain 1
- myomaker, myoblast fusion factor
- myosin XVIIIB
- myosin IXA
- myotilin
- myopalladin
- N-acetyl-alpha-glucosaminidase
- N-myc downstream regulated 1
- 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
- neurofilament light chain
- NIMA related kinase 1
- nerve growth factor
- NIPA magnesium transporter 1
- 5'-nucleotidase, cytosolic II
- neurotrophic receptor tyrosine kinase 1
- NUBP iron-sulfur cluster assembly factor, mitochondrial
- outer mitochondrial membrane lipid metabolism regulator OPA3
- optineurin
- ORAI calcium release-activated calcium modulator 1
- prolyl 4-hydroxylase subunit alpha 1
- poly(A) binding protein nuclear 1
- pantothenate kinase 2
- pyruvate dehydrogenase E1 subunit alpha 1

- pyruvate dehydrogenase kinase 3
- prodynorphin
- PET100 cytochrome c oxidase chaperone
- peroxisomal biogenesis factor 1
- peroxisomal biogenesis factor 7
- phosphofructokinase, muscle
- profilin 1
- phosphoglycerate mutase 2
- post-GPI attachment to proteins inositol deacylase 1
- phosphoglycerate kinase 1
- phosphoglucomutase 1
- phosphorylase kinase regulatory subunit alpha 1
- phosphorylase kinase regulatory subunit beta
- paired like homeobox 2A
- phytanoyl-CoA 2-hydroxylase
- piezo type mechanosensitive ion channel component 2
- phosphatidylinositol-4-phosphate 5-kinase type 1 gamma
- phospholipase A2 group VI
- plectin
- pleckstrin homology and RhoGEF domain containing G5
- proteolipid protein 1
- peripheral myelin protein 2
- peripheral myelin protein 22
- polynucleotide kinase 3'-phosphatase
- patatin like phospholipase domain containing 2
- patatin like phospholipase domain containing 6
- patatin like phospholipase domain containing 8
- protein O-glucosyltransferase 1
- DNA polymerase gamma, catalytic subunit
- DNA polymerase gamma 2, accessory subunit
- RNA polymerase III subunit A
- RNA polymerase III subunit B
- 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
- protein phosphatase 2 regulatory subunit Bbeta
- PR/SET domain 12
- prolyl endopeptidase like
- protein kinase AMP-activated non-catalytic subunit gamma 2
- prion protein
- peripherin
- phosphoribosyl pyrophosphate synthetase 1
- prune exopolyphosphatase 1
- periaxin
- presenilin 1
- peptidyl-tRNA hydrolase 2
- pseudouridine synthase 1
- glycogen phosphorylase, muscle associated
- pyridine nucleotide-disulphide oxidoreductase domain 1
- RAB3 GTPase activating non-catalytic protein subunit 2
- RAB7A, member RAS oncogene family
- receptor associated protein of the synapse
- RANBP2-type and C3HC4-type zinc finger containing 1
- RNA binding motif protein 7
- receptor accessory protein 1
- receptor accessory protein 2
- reticulophagy regulator 1
- ribonuclease H2 subunit B
- ribonuclease T2
- ribonucleotide reductase regulatory TP53 inducible subunit M2B
- reticulon 2
- ribitol xylosyltransferase 1
- ryanodine receptor 1
- ryanodine receptor 3

- [sacsin molecular chaperone](#)
- [SET binding factor 1](#)
- [SET binding factor 2](#)
- [sodium voltage-gated channel alpha subunit 10](#)
- [sodium voltage-gated channel alpha subunit 11](#)
- [sodium voltage-gated channel alpha subunit 4](#)
- [sodium voltage-gated channel alpha subunit 5](#)
- [sodium voltage-gated channel alpha subunit 9](#)
- [synthesis of cytochrome C oxidase 1](#)
- [synthesis of cytochrome C oxidase 2](#)
- [selenoprotein N](#)
- [septin 9](#)
- [senataxin](#)
- [sarcoglycan alpha](#)
- [sarcoglycan beta](#)
- [sarcoglycan delta](#)
- [sarcoglycan epsilon](#)
- [sarcoglycan gamma](#)
- [sphingosine-1-phosphate lyase 1](#)
- [SH3 domain and tetratricopeptide repeats 2](#)
- [sigma non-opioid intracellular receptor 1](#)
- [SIL1 nucleotide exchange factor](#)
- [solute carrier family 12 member 6](#)
- [solute carrier family 16 member 1](#)
- [solute carrier family 16 member 2](#)
- [solute carrier family 18 member A3](#)
- [solute carrier family 22 member 5](#)
- [solute carrier family 25 member 15](#)
- [solute carrier family 25 member 20](#)
- [solute carrier family 25 member 4](#)
- [solute carrier family 25 member 42](#)
- [solute carrier family 25 member 46](#)
- [solute carrier family 2 member 1](#)

- solute carrier family 33 member 1
- 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
- SRY-box transcription factor 10
- spartin
- spastin
- striated muscle enriched protein kinase
- SPG11 vesicle trafficking associated, spatacin
- SPG21 abhydrolase domain containing, maspardin
- SPG7 matrix AAA peptidase subunit, paraplegin
- sepiapterin reductase
- spectrin beta, non-erythrocytic 4
- serine palmitoyltransferase long chain base subunit 1
- serine palmitoyltransferase long chain base subunit 2
- sequestosome 1
- SRSF protein kinase 3
- 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
- Sad1 and UNC84 domain containing 1
- 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
- transport and golgi organization 2 homolog
- TAR DNA binding protein

- threonyl-tRNA synthetase 2, mitochondrial
- tubulin folding cofactor E
- TANK binding kinase 1
- titin-cap
- tyrosyl-DNA phosphodiesterase 1
- tectonin beta-propeller repeat containing 2
- trafficking from ER to golgi regulator
- transglutaminase 6
- tyrosine hydroxylase
- TIA1 cytotoxic granule associated RNA binding protein
- thymidine kinase 2
- transmembrane protein 43
- transmembrane protein 65
- troponin I2, fast skeletal type
- troponin T1, slow skeletal type
- troponin T3, fast skeletal type
- transportin 3
- torsin family 1 member A
- torsin 1A interacting protein 1
- triosephosphate isomerase 1
- tropomyosin 2
- tropomyosin 3
- trafficking protein particle complex subunit 11
- tripartite motif containing 2
- 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
- tau tubulin kinase 2
- titin

- transthyretin
- tubulin alpha 4a
- tubulin beta 3 class III
- tubulin beta 4A class IVa
- twinkle mtDNA helicase
- thymidine phosphorylase
- ubiquitin like modifier activating enzyme 1
- ubiquilin 2
- unc-13 homolog A
- ubiquitin specific peptidase 8
- vesicle associated membrane protein 1
- VAMP associated protein B and C
- valyl-tRNA synthetase 2, mitochondrial
- valosin containing protein
- vacuolar ATPase assembly factor VMA21
- VPS33B late endosome and lysosome associated
- VPS37A subunit of ESCRT-I
- VRK serine/threonine kinase 1
- tryptophanyl-tRNA synthetase 1
- WASH complex subunit 5
- WD repeat domain 45
- WD repeat domain 48
- WNK lysine deficient protein kinase 1
- WW domain containing transcription regulator 1
- X-linked Kx blood group antigen, Kell and VPS13A binding protein
- tyrosyl-tRNA synthetase 1
- tyrosyl-tRNA synthetase 2
- zinc finger C4H2-type containing
- zinc finger homeobox 2
- zinc finger RNA binding protein
- zinc finger FYVE-type containing 26
- zinc finger FYVE-type containing 27

Related Gene Panels

- [Neuromuscular disorders \(548 genes\) - ULB](#)

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