

GENETIC TEST:

Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)

FULL NAME:	Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)
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
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	22 Aug 2019 - 11:00
CHANGED:	17 Aug 2023 - 11:20
URL:	https://ulbgenetics.be/compendium-des-analyses/#GM61

Source URL: http://gentest.healthdata.be/index.php/index.php/genetic_test/591

RELATED CONTENT

Related Laboratories

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

Related Analytes

- [alpha-2-macroglobulin like 1](#)
- [alpha and gamma adaptin binding protein](#)
- [ATP binding cassette subfamily A member 12](#)
- [ATP binding cassette subfamily B member 6 \(Langereis blood group\)](#)
- [ATP binding cassette subfamily C member 6](#)
- [abhydrolase domain containing 5, lysophosphatidic acid acyltransferase](#)
- [ACD shelterin complex subunit and telomerase recruitment factor](#)
- [ADAM metallopeptidase domain 10](#)
- [ADAM metallopeptidase with thrombospondin type 1 motif 2](#)
- [adenosine deaminase RNA specific](#)
- [aurora kinase B](#)
- [AKT serine/threonine kinase 1](#)
- [aldehyde dehydrogenase 18 family member A1](#)
- [aldehyde dehydrogenase 3 family member A2](#)
- [arachidonate 12-lipoxygenase, 12R type](#)
- [arachidonate lipoxygenase 3](#)
- [ANTXR cell adhesion molecule 1](#)
- [adaptor related protein complex 1 subunit sigma 1](#)
- [adaptor related protein complex 3 subunit beta 1](#)
- [APC down-regulated 1](#)

- aquaporin 5
- Rho GTPase activating protein 31
- arylsulfatase L
- N-acylsphingosine amidohydrolase 1
- ATM serine/threonine kinase
- ATPase sarcoplasmic/endoplasmic reticulum Ca²⁺ transporting 2
- ATPase secretory pathway Ca²⁺ transporting 1
- ATPase H⁺ transporting V0 subunit a2
- ATPase H⁺ transporting V1 subunit A
- ATPase H⁺ transporting V1 subunit E1
- ATPase copper transporting alpha
- axin 2
- beta-1,3-galactosyltransferase 6
- beta-1,4-galactosyltransferase 7
- BAF nuclear assembly factor 1
- BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone
- BLM RecQ like helicase
- biogenesis of lysosomal organelles complex 1 subunit 3
- biogenesis of lysosomal organelles complex 1 subunit 6
- B-Raf proto-oncogene, serine/threonine kinase
- caspase recruitment domain family member 14
- Cbl proto-oncogene
- cystathionine beta-synthase
- CD151 molecule (Raph blood group)
- cadherin 3
- cyclin dependent kinase 4
- cyclin dependent kinase inhibitor 1B
- cyclin dependent kinase inhibitor 2A
- corneodesmosin
- ceramide synthase 3
- carbohydrate sulfotransferase 14
- carbohydrate sulfotransferase 8
- component of inhibitor of nuclear factor kappa B kinase complex

- cardiotrophin like cytokine factor 1
- claudin 1
- claudin 10
- component of oligomeric golgi complex 6
- collagen type XI alpha 1 chain
- collagen type XII alpha 1 chain
- collagen type XVII alpha 1 chain
- collagen type I alpha 1 chain
- collagen type I alpha 2 chain
- collagen type III alpha 1 chain
- collagen type V alpha 1 chain
- collagen type V alpha 2 chain
- collagen type VII alpha 1 chain
- CREB binding protein
- cytokine receptor like factor 1
- cystatin A
- CST telomere replication complex component 1
- cathepsin C
- CYLD lysine 63 deubiquitinase
- cytochrome P450 family 26 subfamily C member 1
- cytochrome P450 family 4 subfamily F member 22
- damage specific DNA binding protein 2
- disco interacting protein 2 homolog B
- dyskerin pseudouridine synthase 1
- delta like canonical Notch ligand 4
- distal-less homeobox 3
- DNA methyltransferase 1
- dedicator of cytokinesis 6
- dolichol kinase
- desmocollin 3
- dermatan sulfate epimerase
- desmoglein 1
- desmoglein 2

- desmoglein 4
- desmoplakin
- dystonin
- dystrobrevin binding protein 1
- extracellular matrix protein 1
- ectodysplasin A
- ectodysplasin A receptor
- EDAR associated via death domain
- endothelin 3
- endothelin receptor type B
- EGF containing fibulin extracellular matrix protein 2
- elastin
- ELOVL fatty acid elongase 4
- elongator complex protein 1
- ectonucleotide pyrophosphatase/phosphodiesterase 1
- EGF domain specific O-linked N-acetylglucosamine transferase
- E1A binding protein p300
- ectopic P-granules 5 autophagy tethering factor
- ERCC excision repair 2, TFIIH core complex helicase subunit
- ERCC excision repair 3, TFIIH core complex helicase subunit
- ERCC excision repair 4, endonuclease catalytic subunit
- ERCC excision repair 5, endonuclease
- ERCC excision repair 6, chromatin remodeling factor
- ERCC excision repair 8, CSA ubiquitin ligase complex subunit
- EvC ciliary complex subunit 1
- EvC ciliary complex subunit 2
- exophilin 5
- coagulation factor XII
- FAM111 trypsin like peptidase B
- family with sequence similarity 83 member G
- fibulin 5
- fibrillin 1
- fibrillin 2

- FERM domain containing kindlin 1
- fibroblast growth factor receptor 1
- fibroblast growth factor receptor 2
- fibroblast growth factor receptor 3
- fumarate hydratase
- FKBP prolyl isomerase 14
- folliculin
- filaggrin 2
- forkhead box N1
- frizzled class receptor 6
- polypeptide N-acetylgalactosaminyltransferase 3
- gigaxonin
- glucosylceramidase beta 1
- gamma-glutamyl carboxylase
- growth hormone receptor
- gap junction protein alpha 1
- gap junction protein beta 2
- gap junction protein beta 3
- gap junction protein beta 4
- gap junction protein beta 6
- galactosidase alpha
- galactosidase beta 1
- GNAS complex locus
- golgin, RAB6 interacting
- G protein-coupled receptor 143
- grainyhead like transcription factor 2
- gelsolin
- general transcription factor IIE subunit 2
- general transcription factor IIH subunit 5
- hepcidin antimicrobial peptide
- holocytochrome c synthase
- histone deacetylase 8
- homeostatic iron regulator

- homogentisate 1,2-dioxygenase
- hedgehog acyltransferase
- hemojuvelin BMP co-receptor
- holocarboxylase synthetase
- homeobox C13
- 15-hydroxyprostaglandin dehydrogenase
- HPS1 biogenesis of lysosomal organelles complex 3 subunit 1
- HPS3 biogenesis of lysosomal organelles complex 2 subunit 1
- HPS4 biogenesis of lysosomal organelles complex 3 subunit 2
- HPS5 biogenesis of lysosomal organelles complex 2 subunit 2
- HPS6 biogenesis of lysosomal organelles complex 2 subunit 3
- HR lysine demethylase and nuclear receptor corepressor
- HRas proto-oncogene, GTPase
- intraflagellar transport 122
- intraflagellar transport 43
- inhibitor of nuclear factor kappa B kinase regulatory subunit gamma
- interleukin 31 receptor A
- insulin receptor
- integrin subunit alpha 3
- integrin subunit alpha 6
- integrin subunit beta 4
- junction plakoglobin
- KN motif and ankyrin repeat domains 2
- potassium voltage-gated channel subfamily H member 1
- potassium channel tetramerization domain containing 1
- keratinocyte differentiation factor 1
- lysine demethylase 6A
- 3-ketodihydrosphingosine reductase
- kinesin family member 1A
- KIT proto-oncogene, receptor tyrosine kinase
- KIT ligand
- klotho
- kelch like family member 24

- killin, p53 regulated DNA replication inhibitor
- lysine methyltransferase 2D
- KRAS proto-oncogene, GTPase
- kringle containing transmembrane protein 1
- keratin 1
- keratin 10
- keratin 14
- keratin 16
- keratin 17
- keratin 2
- keratin 5
- keratin 6A
- keratin 6B
- keratin 6C
- keratin 71
- keratin 74
- keratin 81
- keratin 83
- keratin 85
- keratin 86
- keratin 9
- laminin subunit alpha 4
- laminin subunit beta 3
- laminin subunit gamma 2
- LEM domain containing 3
- lipase H
- lipase family member N
- lamin A/C
- LIM homeobox transcription factor 1 beta
- loricrin cornified envelope precursor protein
- lysophosphatidic acid receptor 6
- leucine rich melanocyte differentiation associated
- LDL receptor related protein 1

- latent transforming growth factor beta binding protein 4
- lysosomal trafficking regulator
- leucine zipper like post translational regulator 1
- mitogen-activated protein kinase kinase 1
- mitogen-activated protein kinase kinase 2
- microtubule associated protein RP/EB family member 2
- membrane bound transcription factor peptidase, site 2
- melanocortin 1 receptor
- menin 1
- melanocyte inducing transcription factor
- mutL homolog 1
- melanophilin
- matrix metallopeptidase 1
- matrix metallopeptidase 2
- mannose-P-dolichol utilization defect 1
- M-phase specific PLK1 interacting protein
- mutS homolog 2
- mutS homolog 6
- methylsterol monooxygenase 1
- msh homeobox 1
- mutY DNA glycosylase
- myosin VA
- nectin cell adhesion molecule 1
- nectin cell adhesion molecule 4
- neurofibromin 1
- NF2, moesin-ezrin-radixin like (MERLIN) tumor suppressor
- NFKB inhibitor alpha
- nerve growth factor
- NHP2 ribonucleoprotein
- NIPA like domain containing 4
- NIPBL cohesin loading factor
- NOP10 ribonucleoprotein
- notch receptor 1

- NRAS proto-oncogene, GTPase
- NAD(P) dependent steroid dehydrogenase-like
- 5'-nucleotidase, cytosolic IIIA
- neurotrophic receptor tyrosine kinase 1
- OCA2 melanosomal transmembrane protein
- OFD1 centriole and centriolar satellite protein
- ORAI calcium release-activated calcium modulator 1
- oncostatin M receptor
- peptidyl arginine deiminase 3
- poly(A)-specific ribonuclease
- paired box 3
- platelet derived growth factor receptor beta
- peroxisomal biogenesis factor 7
- phytanoyl-CoA 2-hydroxylase
- phosphatidylinositol glycan anchor biosynthesis class L
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha
- plakophilin 1
- phospholipase C delta 1
- plectin
- procollagen-lysine,2-oxoglutarate 5-dioxygenase 1
- procollagen-lysine,2-oxoglutarate 5-dioxygenase 3
- PMS1 homolog 2, mismatch repair system component
- patatin like phospholipase domain containing 1
- protein O-fucosyltransferase 1
- protein O-glucosyltransferase 1
- DNA polymerase delta 1, catalytic subunit
- DNA polymerase eta
- porcupine O-acyltransferase
- protein phosphatase 1 catalytic subunit beta
- PR/SET domain 12
- PR/SET domain 5
- protein kinase cAMP-dependent type I regulatory subunit alpha
- protein kinase D1

- phosphoserine aminotransferase 1
- presenilin enhancer, gamma-secretase subunit
- patched 1
- patched 2
- phosphatidylserine synthase 1
- phosphatase and tensin homolog
- protein tyrosine phosphatase non-receptor type 11
- pyrroline-5-carboxylate reductase 1
- RAB27A, member RAS oncogene family
- RAD21 cohesin complex component
- Raf-1 proto-oncogene, serine/threonine kinase
- RAS p21 protein activator 2
- recombination signal binding protein for immunoglobulin kappa J region
- RecQ like helicase 4
- ret proto-oncogene
- reticulophagy regulator 1
- rhomboid 5 homolog 2
- Ras and Rab interactor 2
- receptor interacting serine/threonine kinase 4
- Ras like without CAAX 1
- RNA component of mitochondrial RNA processing endoribonuclease
- ring finger protein 113A
- ribosomal protein L21
- RAS related
- R-spondin 1
- regulator of telomere elongation helicase 1
- SAM and SH3 domain containing 1
- sodium voltage-gated channel alpha subunit 11
- sodium voltage-gated channel alpha subunit 9
- succinate dehydrogenase complex iron sulfur subunit B
- succinate dehydrogenase complex subunit D
- SEC23 homolog B, COPII coat complex component
- serpin family B member 7

- [serpin family B member 8](#)
- [serpin family G member 1](#)
- [SET binding protein 1](#)
- [sphingosine-1-phosphate lyase 1](#)
- [SHOC2 leucine rich repeat scaffold protein](#)
- [SKI2 subunit of superkiller complex](#)
- [SKI3 subunit of superkiller complex](#)
- [solute carrier family 24 member 5](#)
- [solute carrier family 27 member 4](#)
- [solute carrier family 29 member 3](#)
- [solute carrier family 2 member 10](#)
- [solute carrier family 39 member 13](#)
- [solute carrier family 39 member 4](#)
- [solute carrier family 45 member 2](#)
- [solute carrier family 6 member 19](#)
- [secreted LY6/PLAUR domain containing 1](#)
- [SMAD family member 3](#)
- [SWI/SNF-related, matrix-associated actin-dependent regulator of chromatin, subfamily a, containing DEAD/H box 1](#)
- [SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1](#)
- [structural maintenance of chromosomes 3](#)
- [sphingomyelin phosphodiesterase 1](#)
- [snail family transcriptional repressor 2](#)
- [synaptosome associated protein 29](#)
- [small nuclear ribonucleoprotein polypeptide E](#)
- [SOS Ras/Rac guanine nucleotide exchange factor 1](#)
- [SOS Ras/Rho guanine nucleotide exchange factor 2](#)
- [SRY-box transcription factor 10](#)
- [serine peptidase inhibitor Kazal type 5](#)
- [sprouty related EVH1 domain containing 1](#)
- [sprouty RTK signaling antagonist 1](#)
- [serine palmitoyltransferase long chain base subunit 1](#)
- [steroid 5 alpha-reductase 3](#)
- [ST14 transmembrane serine protease matriptase](#)

- serine/threonine kinase 11
- steroid sulfatase
- SUFU negative regulator of hedgehog signaling
- sulfotransferase family 2B member 1
- sulfatase modifying factor 1
- tyrosine aminotransferase
- trichohyalin
- telomerase RNA component
- telomerase reverse transcriptase
- transferrin receptor 2
- transforming growth factor beta 2
- transforming growth factor beta 3
- transforming growth factor beta receptor 1
- transforming growth factor beta receptor 2
- transglutaminase 1
- transglutaminase 3
- transglutaminase 5
- TERF1 interacting nuclear factor 2
- transmembrane channel like 6
- tenascin XB
- tumor protein p63
- transcriptional repressor GATA binding 1
- transient receptor potential cation channel subfamily V member 3
- TSC complex subunit 1
- TSC complex subunit 2
- tubulin beta class I
- twist family bHLH transcription factor 2
- tyrosinase
- tyrosinase related protein 1
- ubiquitin protein ligase E3 component n-recognin 1
- U6 snRNA biogenesis phosphodiesterase 1
- UV stimulated scaffold protein A
- VPS33B late endosome and lysosome associated

- [WD repeat domain 19](#)
- [WD repeat domain 35](#)
- [WNK lysine deficient protein kinase 1](#)
- [Wnt family member 10A](#)
- [WD repeat containing antisense to TP53](#)
- [WRN RecQ like helicase](#)
- [XPA, DNA damage recognition and repair factor](#)
- [XPC complex subunit, DNA damage recognition and repair factor](#)
- [zinc metallopeptidase STE24](#)
- [zinc finger protein 469](#)
- [zinc finger protein 750](#)

Related Gene Panels

- [Dermatogenetic / severe, rare and hereditary genodermatoses \(394 genes\) - ULB](#)

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