

GENETIC TEST:

Primary immune deficiencies (gene panel)

FULL NAME:	Primary immune deficiencies (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
EQA:	• Primary Immunodeficiencies
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	01 Aug 2019 - 11:44

CHANGED:	16 Dec 2022 - 13:03
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Source URL: http://gentest.healthdata.be/genetic_test/350

RELATED CONTENT

Related Laboratories

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

Related Analytes

- [ACD shelterin complex subunit and telomerase recruitment factor](#)
- [acid phosphatase 5, tartrate resistant](#)
- [actin beta](#)
- [adenosine deaminase](#)
- [adenosine deaminase 2](#)
- [ADAM metallopeptidase domain 17](#)
- [adenosine deaminase RNA specific](#)
- [activation induced cytidine deaminase](#)
- [autoimmune regulator](#)
- [adenylate kinase 2](#)
- [alkaline phosphatase, intestinal](#)
- [adaptor related protein complex 1 subunit sigma 3](#)
- [adaptor related protein complex 3 subunit beta 1](#)
- [adaptor related protein complex 3 subunit delta 1](#)
- [apolipoprotein L1](#)
- [Rho guanine nucleotide exchange factor 1](#)
- [actin related protein 2/3 complex subunit 1B](#)
- [autophagy related 16 like 1](#)
- [autophagy related 4A cysteine peptidase](#)
- [ATM serine/threonine kinase](#)

- ATPase sarcoplasmic/endoplasmic reticulum Ca²⁺ transporting 2
- ATPase H⁺ transporting accessory protein 1
- beta-2-microglobulin
- BTB domain and CNC homolog 2
- BCL10 immune signaling adaptor
- BAF chromatin remodeling complex subunit BCL11B
- BLK proto-oncogene, Src family tyrosine kinase
- BLM RecQ like helicase
- B cell linker
- biogenesis of lysosomal organelles complex 1 subunit 3
- biogenesis of lysosomal organelles complex 1 subunit 6
- Bruton tyrosine kinase
- complement C1q A chain
- complement C1q B chain
- complement C1q C chain
- complement C1r
- complement C1s
- complement C2
- chromosome 2 open reading frame 69
- complement C3
- complement C4A (Rodgers blood group)
- complement component 4 binding protein alpha
- complement C5
- complement C6
- complement C7
- complement C8 alpha chain
- complement C8 beta chain
- complement C8 gamma chain
- complement C9
- caspase recruitment domain family member 11
- caspase recruitment domain family member 14
- caspase recruitment domain family member 9
- capping protein regulator and myosin 1 linker 2

- caspase 10
- caspase 8
- Cbl proto-oncogene
- collagen and calcium binding EGF domains 1
- coiled-coil domain containing 28B
- CD19 molecule
- CD247 molecule
- CD27 molecule
- CD3 delta subunit of T-cell receptor complex
- CD3 epsilon subunit of T-cell receptor complex
- CD3 gamma subunit of T-cell receptor complex
- CD4 molecule
- CD40 molecule
- CD40 ligand
- CD46 molecule
- CD55 molecule (Cromer blood group)
- CD59 molecule (CD59 blood group)
- CD70 molecule
- CD79a molecule
- CD79b molecule
- CD81 molecule
- CD8 subunit alpha
- cell division cycle 42
- cell division cycle associated 7
- CCAAT enhancer binding protein epsilon
- complement factor B
- complement factor D
- complement factor H
- complement factor H related 1
- complement factor H related 2
- complement factor H related 3
- complement factor H related 4
- complement factor H related 5

- complement factor I
- complement factor properdin
- CF transmembrane conductance regulator
- chromodomain helicase DNA binding protein 7
- calcium and integrin binding 1
- class II major histocompatibility complex transactivator
- chloride voltage-gated channel 7
- C-type lectin domain containing 7A
- ClpB family mitochondrial disaggregase
- collagen type VII alpha 1 chain
- COPI coat complex subunit alpha
- coronin 1A
- carnitine palmitoyltransferase 2
- complement C3d receptor 2
- CREB binding protein
- colony stimulating factor 2 receptor subunit alpha
- colony stimulating factor 2 receptor subunit beta
- colony stimulating factor 3 receptor
- CST telomere replication complex component 1
- cytotoxic T-lymphocyte associated protein 4
- catenin beta like 1
- CTP synthase 1
- cathepsin C
- C-X-C motif chemokine receptor 4
- cytochrome b-245 alpha chain
- cytochrome b-245 beta chain
- cytochrome b-245 chaperone 1
- debranching RNA lariats 1
- DNA cross-link repair 1B
- DNA cross-link repair 1C
- DEF6 guanine nucleotide exchange factor
- diacylglycerol kinase epsilon
- dihydrofolate reductase

- dyskerin pseudouridine synthase 1
- DnaJ heat shock protein family (Hsp40) member C21
- deoxyribonuclease 1
- deoxyribonuclease 1 like 3
- deoxyribonuclease 2, lysosomal
- DNA methyltransferase 3 beta
- dedicator of cytokinesis 2
- dedicator of cytokinesis 8
- docking protein 3
- dystrobrevin binding protein 1
- elongation factor like GTPase 1
- elastase, neutrophil expressed
- E74 like ETS transcription factor 4
- ectopic P-granules 5 autophagy tethering factor
- erbb2 interacting protein
- ERCC excision repair 6 like 2
- exostosin like glycosyltransferase 3
- coagulation factor XII
- FA core complex associated protein 24
- Fas associated via death domain
- Fas cell surface death receptor
- Fas ligand
- FAT atypical cadherin 4
- Fc fragment of IgG receptor IIb
- Fc fragment of IgG receptor IIIa
- Fc fragment of IgG receptor IIIb
- FCH and mu domain containing endocytic adaptor 1
- ficolin 3
- FERM domain containing kindlin 1
- FERM domain containing kindlin 3
- folliculin interacting protein 1
- forkhead box N1
- forkhead box P3

- formyl peptide receptor 1
- glucose-6-phosphatase catalytic subunit 3
- glucose-6-phosphate dehydrogenase
- GATA binding protein 1
- GATA binding protein 2
- growth factor independent 1 transcriptional repressor
- GTPase, IMAP family member 5
- GIN5 complex subunit 1
- guanylate cyclase 2C
- hepatitis A virus cellular receptor 2
- HCLS1 associated protein X-1
- helicase, lymphoid specific
- heme oxygenase 1
- HPS1 biogenesis of lysosomal organelles complex 3 subunit 1
- HPS4 biogenesis of lysosomal organelles complex 3 subunit 2
- HPS6 biogenesis of lysosomal organelles complex 2 subunit 3
- HtrA serine peptidase 2
- hypoxia up-regulated 1
- inducible T cell costimulator
- inducible T cell costimulator ligand
- interferon induced with helicase C domain 1
- interferon alpha and beta receptor subunit 1
- interferon alpha and beta receptor subunit 2
- interferon gamma
- interferon gamma receptor 1
- interferon gamma receptor 2
- immunoglobulin heavy constant mu
- immunoglobulin kappa constant
- immunoglobulin lambda like polypeptide 1
- inhibitor of nuclear factor kappa B kinase subunit beta
- inhibitor of nuclear factor kappa B kinase regulatory subunit gamma
- IKAROS family zinc finger 1
- interleukin 10

- interleukin 10 receptor subunit alpha
- interleukin 10 receptor subunit beta
- interleukin 12B
- interleukin 12 receptor subunit beta 1
- interleukin 12 receptor subunit beta 2
- interleukin 15 receptor subunit alpha
- interleukin 17F
- interleukin 17 receptor A
- interleukin 17 receptor C
- interleukin 18
- interleukin 18 binding protein
- interleukin 1 receptor like 1
- interleukin 1 receptor antagonist
- interleukin 21
- interleukin 21 receptor
- interleukin 23 receptor
- interleukin 2 receptor subunit alpha
- interleukin 2 receptor subunit beta
- interleukin 2 receptor subunit gamma
- interleukin 36 receptor antagonist
- interleukin 6 receptor
- interleukin 6 cytokine family signal transducer
- interleukin 7 receptor
- inflammation and lipid regulator with UBA-like and NBR1-like domains
- INO80 complex ATPase subunit
- interleukin 1 receptor associated kinase 1
- interleukin 1 receptor associated kinase 4
- interferon regulatory factor 2 binding protein 2
- interferon regulatory factor 3
- interferon regulatory factor 4
- interferon regulatory factor 7
- interferon regulatory factor 8
- interferon regulatory factor 9

- ISG15 ubiquitin like modifier
- itchy E3 ubiquitin protein ligase
- integrin subunit beta 2
- IL2 inducible T cell kinase
- inositol-trisphosphate 3-kinase B
- influenza virus NS1A binding protein
- jagunal homolog 1
- Janus kinase 1
- Janus kinase 3
- lysine demethylase 6A
- lysine methyltransferase 2A
- lysine methyltransferase 2D
- KRAS proto-oncogene, GTPase
- laccase domain containing 1
- late endosomal/lysosomal adaptor, MAPK and MTOR activator 2
- linker for activation of T cells
- LCK proto-oncogene, Src family tyrosine kinase
- lymphocyte cytosolic protein 2
- DNA ligase 1
- DNA ligase 4
- lipase A, lysosomal acid type
- lipin 2
- LPS responsive beige-like anchor protein
- leucine rich repeat containing 8 VRAC subunit A
- LSM11, U7 small nuclear RNA associated
- lysosomal trafficking regulator
- magnesium transporter 1
- MALT1 paracaspase
- mannosidase alpha class 2B member 1
- microtubule associated protein 1 light chain 3 beta 2
- mitogen-activated protein kinase kinase kinase 14
- mitogen-activated protein kinase 8
- MBL associated serine protease 1

- MBL associated serine protease 2
- mannose binding lectin 2
- minichromosome maintenance 10 replication initiation factor
- minichromosome maintenance complex component 4
- MEFV innate immunity regulator, pyrin
- mannosyl-oligosaccharide glucosidase
- macrophage expressed 1
- myeloperoxidase
- MRE11 homolog, double strand break repair nuclease
- myocardin related transcription factor A
- membrane spanning 4-domains A1
- mutS homolog 6
- moesin
- methylenetetrahydrofolate dehydrogenase, cyclohydrolase and formyltetrahydrofolate synthetase 1
- mevalonate kinase
- MYD88 innate immune signal transduction adaptor
- myosin VB
- Myb like, SWIRM and MPN domains 1
- NBAS subunit of NRZ tethering complex
- nibrin
- neutrophil cytosolic factor 1
- neutrophil cytosolic factor 2
- neutrophil cytosolic factor 4
- NCK associated protein 1 like
- nicastatin
- nuclear factor of activated T cells 5
- NFE2 like bZIP transcription factor 2
- nuclear factor kappa B subunit 1
- nuclear factor kappa B subunit 2
- NFkB inhibitor alpha
- non-homologous end joining factor 1
- NHP2 ribonucleoprotein
- NK2 homeobox 5

- NLR family CARD domain containing 4
- NLR family pyrin domain containing 1
- NLR family pyrin domain containing 12
- NLR family pyrin domain containing 3
- NLR family pyrin domain containing 7
- nucleotide binding oligomerization domain containing 2
- NOP10 ribonucleoprotein
- nitric oxide synthase 2
- NPC intracellular cholesterol transporter 1
- NRAS proto-oncogene, GTPase
- NSE3 homolog, SMC5-SMC6 complex component
- 5'-nucleotidase, cytosolic IIIA
- 2'-5'-oligoadenylate synthetase 1
- ORAI calcium release-activated calcium modulator 1
- osteoclastogenesis associated transmembrane protein 1
- OTU deubiquitinase with linear linkage specificity
- poly(A)-specific ribonuclease
- paired box 1
- propionyl-CoA carboxylase subunit alpha
- propionyl-CoA carboxylase subunit beta
- peptidase D
- phosphoglucomutase 3
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit delta
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit gamma
- phosphoinositide-3-kinase regulatory subunit 1
- phospholipase C gamma 2
- pleckstrin homology and RUN domain containing M1
- plasminogen
- peripheral myelin protein 22
- PMS1 homolog 2, mismatch repair system component
- purine nucleoside phosphorylase
- DNA polymerase alpha 1, catalytic subunit
- DNA polymerase delta 1, catalytic subunit

- DNA polymerase delta 2, accessory subunit
- DNA polymerase epsilon, catalytic subunit
- DNA polymerase epsilon 2, accessory subunit
- RNA polymerase III subunit A
- RNA polymerase III subunit C
- RNA polymerase III subunit F
- perforin 1
- DNA primase subunit 1
- protein kinase C delta
- protein kinase, DNA-activated, catalytic subunit
- presenilin 1
- presenilin enhancer, gamma-secretase subunit
- proteasome 20S subunit alpha 3
- proteasome 20S subunit beta 10
- proteasome 20S subunit beta 4
- proteasome 20S subunit beta 8
- proteasome 20S subunit beta 9
- proteasome assembly chaperone 2
- proline-serine-threonine phosphatase interacting protein 1
- proline-serine-threonine phosphatase interacting protein 2
- phosphatase and tensin homolog
- protein tyrosine phosphatase non-receptor type 11
- protein tyrosine phosphatase non-receptor type 2
- protein tyrosine phosphatase non-receptor type 6
- protein tyrosine phosphatase receptor type C
- RAB27A, member RAS oncogene family
- Rac family small GTPase 2
- recombination activating 1
- recombination activating 2
- RAN binding protein 2
- RAS guanyl releasing protein 1
- RANBP2-type and C3HC4-type zinc finger containing 1
- ring finger and CCCH-type domains 1

- RecQ like helicase 4
- REL proto-oncogene, NF-kB subunit
- RELA proto-oncogene, NF-kB subunit
- RELB proto-oncogene, NF-kB subunit
- regulatory factor X5
- regulatory factor X associated ankyrin containing protein
- regulatory factor X associated protein
- ras homolog family member H
- RNA sensor RIG-I
- receptor interacting serine/threonine kinase 1
- RNA component of mitochondrial RNA processing endoribonuclease
- ribonuclease H2 subunit A
- ribonuclease H2 subunit B
- ribonuclease H2 subunit C
- ring finger protein 168
- ring finger protein 31
- RAR related orphan receptor C
- ribosomal protein SA
- regulator of telomere elongation helicase 1
- sterile alpha motif domain containing 9
- sterile alpha motif domain containing 9 like
- SAM and HD domain containing deoxynucleoside triphosphate triphosphohydrolase 1
- SAM and SH3 domain containing 3
- SBDS ribosome maturation factor
- succinate dehydrogenase complex flavoprotein subunit A
- SEC61 translocon subunit alpha 1
- semaphorin 3E
- serpin family G member 1
- sphingosine-1-phosphate lyase 1
- SH2 domain containing 1A
- SH3 domain binding protein 2
- SH3 domain containing kinase binding protein 1
- SKI2 subunit of superkiller complex

- SKI3 subunit of superkiller complex
- solute carrier family 11 member 1
- solute carrier family 29 member 3
- solute carrier family 35 member C1
- solute carrier family 37 member 4
- solute carrier family 39 member 7
- solute carrier family 46 member 1
- solute carrier family 7 member 7
- solute carrier family 9 member A3
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a like 1
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily d, member 2
- sorting nexin 10
- suppressor of cytokine signaling 1
- suppressor of cytokine signaling 4
- Spi-1 proto-oncogene
- serine peptidase inhibitor Kazal type 5
- signal peptide peptidase like 2A
- signal recognition particle 54
- signal recognition particle 72
- signal transducer and activator of transcription 1
- signal transducer and activator of transcription 2
- signal transducer and activator of transcription 3
- signal transducer and activator of transcription 4
- signal transducer and activator of transcription 5B
- signal transducer and activator of transcription 6
- stromal interaction molecule 1
- stimulator of interferon response cGAMP interactor 1
- serine/threonine kinase 4
- STN1 subunit of CST complex
- syntaxin 11
- syntaxin binding protein 2
- syntaxin binding protein 3
- spleen associated tyrosine kinase

- tafazzin, phospholipid-lysophospholipid transacylase
- transporter 1, ATP binding cassette subfamily B member
- transporter 2, ATP binding cassette subfamily B member
- TAP binding protein
- TANK binding kinase 1
- T-box transcription factor 1
- T-box transcription factor 21
- transcription factor 3
- T cell immune regulator 1, ATPase H⁺ transporting V0 subunit a3
- transcobalamin 2
- telomerase RNA component
- telomerase reverse transcriptase
- tet methylcytosine dioxygenase 2
- transferrin receptor
- transforming growth factor beta 1
- transforming growth factor beta receptor 1
- transforming growth factor beta receptor 2
- thrombomodulin
- TIR domain containing adaptor molecule 1
- TERF1 interacting nuclear factor 2
- TIR domain containing adaptor protein
- toll like receptor 3
- toll like receptor 4
- toll like receptor 7
- transmembrane channel like 6
- transmembrane channel like 8
- TNF alpha induced protein 3
- TNF receptor superfamily member 11a
- TNF receptor superfamily member 13B
- TNF receptor superfamily member 13C
- TNF receptor superfamily member 1A
- TNF receptor superfamily member 4
- TNF receptor superfamily member 9

- TNF superfamily member 11
- TNF superfamily member 12
- TNF superfamily member 13
- DNA topoisomerase II beta
- tripeptidyl peptidase 2
- T-cell receptor alpha constant
- TNF receptor associated factor 3
- TRAF3 interacting protein 2
- three prime repair exonuclease 1
- tripartite motif containing 22
- tRNA nucleotidyl transferase 1
- tetratricopeptide repeat domain 7A
- tyrosine kinase 2
- ubiquitin like modifier activating enzyme 1
- unc-119 lipid binding chaperone
- unc-13 homolog D
- unc-93 homolog B1, TLR signaling regulator
- uracil DNA glycosylase
- U6 snRNA biogenesis phosphodiesterase 1
- ubiquitin specific peptidase 18
- vav guanine nucleotide exchange factor 1
- vacuolar protein sorting 13 homolog B
- vacuolar protein sorting 45 homolog
- WASP actin nucleation promoting factor
- WD repeat domain 1
- WAS/WASL interacting protein family member 1
- WD repeat containing antisense to TP53
- X-box binding protein 1
- X-linked inhibitor of apoptosis
- zeta chain of T-cell receptor associated protein kinase 70
- zinc finger and BTB domain containing 24
- zinc finger protein 341
- zinc finger NFX1-type containing 1

Related Gene Panels

- [Primary immune deficiencies - UGent](#)

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