

GENETIC TEST: Skeletal dysplasia (gene panel)

FULL NAME:	Skeletal dysplasia (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 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
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	15 Dec 2020 - 17:00
CHANGED:	16 Dec 2022 - 13:18

Source URL: http://gentest.healthdata.be/genetic_test/1037

RELATED CONTENT

Related Laboratories

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

Related Analytes

- [ATP binding cassette subfamily C member 9](#)
- [ABL proto-oncogene 1, non-receptor tyrosine kinase](#)
- [aggrecan](#)
- [acid phosphatase 5, tartrate resistant](#)
- [activin A receptor type 1](#)
- [ADAM metallopeptidase with thrombospondin type 1 motif 10](#)
- [ADAM metallopeptidase with thrombospondin type 1 motif 17](#)
- [ADAMTS like 2](#)
- [ALF transcription elongation factor 4](#)
- [aspartylglucosaminidase](#)
- [alkylglycerone phosphate synthase](#)
- [AT-hook DNA binding motif containing 1](#)
- [AKT serine/threonine kinase 1](#)
- [ALG12 alpha-1,6-mannosyltransferase](#)
- [ALG3 alpha-1,3- mannosyltransferase](#)
- [ALG9 alpha-1,2-mannosyltransferase](#)
- [alkaline phosphatase, biomineralization associated](#)
- [ALX homeobox 1](#)
- [ALX homeobox 3](#)
- [ALX homeobox 4](#)

- APC membrane recruitment protein 1
- anaphase promoting complex subunit 1
- ANKH inorganic pyrophosphate transport regulator
- ankyrin repeat domain containing 11
- anoctamin 5
- ANTXR cell adhesion molecule 2
- Rho GTPase activating protein 31
- AT-rich interaction domain 1A
- AT-rich interaction domain 1B
- arylsulfatase L
- ASXL transcriptional regulator 1
- ASXL transcriptional regulator 2
- ATPase H⁺ transporting V0 subunit a2
- beta-1,3-galactosyltransferase 6
- beta-1,3-glucuronyltransferase 3
- beta 3-glucosyltransferase
- beta-1,4-galactosyltransferase 7
- B9 domain containing 1
- Bardet-Biedl syndrome 1
- Bardet-Biedl syndrome 10
- Bardet-Biedl syndrome 12
- Bardet-Biedl syndrome 2
- Bardet-Biedl syndrome 4
- Bardet-Biedl syndrome 5
- Bardet-Biedl syndrome 7
- Bardet-Biedl syndrome 9
- basic helix-loop-helix family member a9
- bone morphogenetic protein 1
- bone morphogenetic protein 2
- BMP binding endothelial regulator
- bone morphogenetic protein receptor type 1B
- 3'(2'), 5'-bisphosphate nucleotidase 2
- blood vessel epicardial substance

- C2 domain containing 3 centriole elongation regulator
- carbonic anhydrase 2
- calcium activated nucleotidase 1
- calcium sensing receptor
- coiled-coil and C2 domain containing 2A
- coiled-coil domain containing 134
- coiled-coil domain containing 8
- cellular communication network factor 6
- cyclin Q
- CD96 molecule
- cell division cycle 45
- cell division cycle 6
- cadherin 3
- cyclin dependent kinase inhibitor 1C
- chromatin licensing and DNA replication factor 1
- centrosomal protein 120
- centrosomal protein 290
- cilia and flagella associated protein 410
- chromodomain helicase DNA binding protein 7
- carbohydrate sulfotransferase 14
- carbohydrate sulfotransferase 3
- chondroitin sulfate synthase 1
- ciliogenesis associated kinase 1
- cytoskeleton associated protein 2 like
- chloride voltage-gated channel 5
- chloride voltage-gated channel 7
- component of oligomeric golgi complex 1
- component of oligomeric golgi complex 4
- collagen type X alpha 1 chain
- collagen type XI alpha 1 chain
- collagen type XI alpha 2 chain
- collagen type I alpha 1 chain
- collagen type I alpha 2 chain

- collagen type II alpha 1 chain
- collagen type IX alpha 1 chain
- collagen type IX alpha 2 chain
- collagen type IX alpha 3 chain
- collectin subfamily member 11
- cartilage oligomeric matrix protein
- corin, serine peptidase
- ciliogenesis and planar polarity effector complex subunit 1
- cAMP responsive element binding protein 3 like 1
- CREB binding protein
- cartilage associated protein
- colony stimulating factor 1 receptor
- chondroitin sulfate N-acetylgalactosaminyltransferase 1
- centrosome and spindle pole associated protein 1
- cathepsin A
- cathepsin C
- cathepsin K
- cullin 7
- cytochrome P450 family 27 subfamily B member 1
- cytochrome P450 family 2 subfamily R member 1
- DCC netrin 1 receptor
- discoidin domain receptor tyrosine kinase 2
- DEAD-box helicase 59
- 24-dehydrocholesterol reductase
- 7-dehydrocholesterol reductase
- dihydroorotate dehydrogenase (quinone)
- disco interacting protein 2 homolog C
- delta like canonical Notch ligand 3
- delta like canonical Notch ligand 4
- distal-less homeobox 3
- distal-less homeobox 5
- distal-less homeobox 6
- dentin matrix acidic phosphoprotein 1

- DNA methyltransferase 3 alpha
- dedicator of cytokinesis 6
- dolichyl-phosphate N-acetylglucosaminophosphotransferase 1
- dolichyl-phosphate mannosyltransferase subunit 1, catalytic
- dentin sialophosphoprotein
- dishevelled segment polarity protein 1
- dishevelled segment polarity protein 3
- dymeclin
- dynein cytoplasmic 2 heavy chain 1
- dynein 2 intermediate chain 1
- dynein 2 intermediate chain 2
- dynein cytoplasmic 2 light intermediate chain 1
- dynein light chain Tctex-type 2B
- endothelin receptor type A
- ephrin A4
- ephrin B1
- elongation factor Tu GTP binding domain containing 2
- eukaryotic translation initiation factor 2 alpha kinase 3
- eukaryotic translation initiation factor 4A3
- ectonucleotide pyrophosphatase/phosphodiesterase 1
- EGF domain specific O-linked N-acetylglucosamine transferase
- E1A binding protein p300
- ERCC excision repair 4, endonuclease catalytic subunit
- ETS2 repressor factor
- establishment of sister chromatid cohesion N-acetyltransferase 2
- EvC ciliary complex subunit 1
- EvC ciliary complex subunit 2
- exostosin glycosyltransferase 1
- exostosin glycosyltransferase 2
- exostosin like glycosyltransferase 3
- enhancer of zeste 2 polycomb repressive complex 2 subunit
- FAM111 trypsin like peptidase A
- FAM20C golgi associated secretory pathway kinase

- fibulin 1
- fibrillin 1
- fibrillin 2
- F-box and WD repeat domain containing 4
- FERM domain containing kindlin 3
- fibroblast growth factor 10
- fibroblast growth factor 16
- fibroblast growth factor 23
- fibroblast growth factor 3
- fibroblast growth factor 4
- fibroblast growth factor 8
- fibroblast growth factor 9
- fibroblast growth factor receptor 1
- fibroblast growth factor receptor 2
- fibroblast growth factor receptor 3
- FIG4 phosphoinositide 5-phosphatase
- FKBP prolyl isomerase 10
- filamin A
- filamin B
- formin 1
- FRAS1 related extracellular matrix 1
- alpha-L-fucosidase 1
- frizzled class receptor 2
- galactosamine (N-acetyl)-6-sulfatase
- polypeptide N-acetylgalactosaminyltransferase 3
- growth differentiation factor 3
- growth differentiation factor 5
- growth differentiation factor 6
- growth factor, augments liver regeneration
- gap junction protein alpha 1
- galactosidase beta 1
- GLI family zinc finger 3
- GNAS complex locus

- glyceronephosphate O-acyltransferase
- glucosamine-phosphate N-acetyltransferase 1
- N-acetylglucosamine-1-phosphate transferase subunits alpha and beta
- N-acetylglucosamine-1-phosphate transferase subunit gamma
- glucosamine (N-acetyl)-6-sulfatase
- golgin, RAB6 interacting
- glypican 6
- glutathione peroxidase 4
- gremlin 1, DAN family BMP antagonist
- glucuronidase beta
- GDNF inducible zinc finger protein 1
- histone deacetylase 4
- histone deacetylase 8
- hes family bHLH transcription factor 7
- heparan-alpha-glucosaminide N-acetyltransferase
- hedgehog acyltransferase
- heterogeneous nuclear ribonucleoprotein K
- homeobox A11
- homeobox A13
- homeobox D13
- 15-hydroxyprostaglandin dehydrogenase
- heparan sulfate 2-O-sulfotransferase 1
- heparan sulfate proteoglycan 2
- isocitrate dehydrogenase (NADP(+)) 1
- isocitrate dehydrogenase (NADP(+)) 2
- iduronate 2-sulfatase
- alpha-L-iduronidase
- interferon induced with helicase C domain 1
- interferon induced transmembrane protein 5
- intraflagellar transport 122
- intraflagellar transport 140
- intraflagellar transport 172
- intraflagellar transport 43

- [intraflagellar transport 52](#)
- [intraflagellar transport 80](#)
- [intraflagellar transport 81](#)
- [Indian hedgehog signaling molecule](#)
- [inhibitor of nuclear factor kappa B kinase regulatory subunit gamma](#)
- [interleukin 11 receptor subunit alpha](#)
- [interleukin 1 receptor antagonist](#)
- [inositol polyphosphate phosphatase like 1](#)
- [lysine acetyltransferase 6B](#)
- [KDEL endoplasmic reticulum protein retention receptor 2](#)
- [KIAA0753](#)
- [KIAA1217](#)
- [kinesin family member 22](#)
- [kinesin family member 7](#)
- [lysine methyltransferase 2D](#)
- [lamin B receptor](#)
- [LEM domain containing 3](#)
- [LFNG O-fucosylpeptide 3-beta-N-acetylglucosaminyltransferase](#)
- [LIF receptor subunit alpha](#)
- [limb development membrane protein 1](#)
- [lamin A/C](#)
- [LIM homeobox transcription factor 1 beta](#)
- [lon peptidase 1, mitochondrial](#)
- [lipin 2](#)
- [LDL receptor related protein 5](#)
- [latent transforming growth factor beta binding protein 2](#)
- [MAF bZIP transcription factor B](#)
- [mannosidase alpha class 2B member 1](#)
- [mannosidase alpha class 2C member 1](#)
- [mitogen-activated protein kinase kinase kinase 7](#)
- [MBL associated serine protease 1](#)
- [matrilin 3](#)
- [membrane bound transcription factor peptidase, site 2](#)

- multiple EGF like domains 8
- mesenchyme homeobox 1
- mesoderm posterior bHLH transcription factor 2
- matrix Gla protein
- MIA SH3 domain ER export factor 2
- MIA SH3 domain ER export factor 3
- MKKS centrosomal shuttling protein
- MKS transition zone complex subunit 1
- matrix metalloproteinase 13
- matrix metalloproteinase 2
- matrix metalloproteinase 9
- motor neuron and pancreas homeobox 1
- mannose-P-dolichol utilization defect 1
- msh homeobox 2
- metaxin 2
- MYCN proto-oncogene, bHLH transcription factor
- myosin light chain 11
- N-acetyl-alpha-glucosaminidase
- N-acetylneuraminase synthase
- NBAS subunit of NRZ tethering complex
- NIMA related kinase 1
- neuraminidase 1
- neurofibromin 1
- nuclear factor I X
- NIPBL cohesin loading factor
- NK3 homeobox 2
- NLR family pyrin domain containing 3
- noggin
- notch receptor 1
- notch receptor 2
- natriuretic peptide C
- NPR2 like, GATOR1 complex subunit
- nuclear receptor binding SET domain protein 1

- NAD(P) dependent steroid dehydrogenase-like
- nucleoredoxin
- obscurin like cytoskeletal adaptor 1
- OFD1 centriole and centriolar satellite protein
- origin recognition complex subunit 4
- origin recognition complex subunit 6
- osteoclastogenesis associated transmembrane protein 1
- prolyl 3-hydroxylase 1
- prolyl 4-hydroxylase subunit beta
- presequence translocase associated motor 16
- poly(A) specific ribonuclease subunit PAN2
- 3'-phosphoadenosine 5'-phosphosulfate synthase 2
- paired box 3
- pericentrin
- phosphate cytidylyltransferase 1A, choline
- phosphodiesterase 3A
- phosphodiesterase 4D
- peroxisomal biogenesis factor 5
- peroxisomal biogenesis factor 7
- phosphoglucomutase 3
- phosphate regulating endopeptidase X-linked
- phosphoglycerate dehydrogenase
- phosphatidylinositol glycan anchor biosynthesis class T
- phosphatidylinositol glycan anchor biosynthesis class V
- phosphatidylinositol-4-phosphate 3-kinase catalytic subunit type 2 alpha
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha
- phosphatidylserine decarboxylase
- paired like homeodomain 1
- pleckstrin homology and RUN domain containing M1
- procollagen-lysine,2-oxoglutarate 5-dioxygenase 2
- plastin 3
- POC1 centriolar protein A
- RNA polymerase I subunit A

- RNA polymerase I and III subunit C
- RNA polymerase I and III subunit D
- porcupine O-acyltransferase
- peptidylprolyl isomerase B
- protein kinase cAMP-dependent type I regulatory subunit alpha
- protein kinase cGMP-dependent 2
- protein arginine methyltransferase 7
- phosphoserine aminotransferase 1
- phosphoserine phosphatase
- phosphatidylserine synthase 1
- parathyroid hormone 1 receptor
- parathyroid hormone like hormone
- protein tyrosine phosphatase non-receptor type 11
- poly(U) binding splicing factor 60
- pyrroline-5-carboxylate reductase 1
- RAB23, member RAS oncogene family
- RAB33B, member RAS oncogene family
- RAD21 cohesin complex component
- RAS guanyl releasing protein 2
- RNA binding motif protein 8A
- recombination signal binding protein for immunoglobulin kappa J region
- RecQ like helicase 4
- RFT1 homolog
- RNA sensor RIG-I
- Ras and Rab interactor 1
- RNA, U4atac small nuclear (U12-dependent splicing)
- receptor tyrosine kinase like orphan receptor 2
- RPGRIP1 like
- ribosomal protein L13
- RUNX family transcription factor 2
- spalt like transcription factor 1
- spalt like transcription factor 4
- SBDS ribosome maturation factor

- scavenger receptor class F member 2
- signal peptide, CUB domain and EGF like domain containing 3
- SEC24 homolog D, COPII coat complex component
- serpin family F member 1
- serpin family H member 1
- SET domain containing 2, histone lysine methyltransferase
- splicing factor 3b subunit 4
- secreted frizzled related protein 4
- N-sulfoglucosamine sulfohydrolase
- SH3 domain binding protein 2
- SH3 and PX domains 2B
- sonic hedgehog signaling molecule
- SHOX homeobox
- solute carrier family 10 member 7
- solute carrier family 17 member 5
- solute carrier family 25 member 15
- solute carrier family 26 member 2
- solute carrier family 29 member 3
- solute carrier family 34 member 1
- solute carrier family 34 member 3
- solute carrier family 35 member C1
- solute carrier family 35 member D1
- solute carrier family 39 member 13
- solute carrier organic anion transporter family member 2A1
- solute carrier organic anion transporter family member 5A1
- secreted LY6/PLAUR domain containing 1
- SMAD family member 3
- SMAD family member 4
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a like 1
- structural maintenance of chromosomes 1A
- structural maintenance of chromosomes 3
- smoothened, frizzled class receptor
- SPARC related modular calcium binding 1

- small nuclear ribonucleoprotein polypeptides B and B1
- sorting nexin 10
- sclerostin
- SRY-box transcription factor 9
- Sp7 transcription factor
- secreted protein acidic and cysteine rich
- SUN domain containing ossification factor
- sulfatase 1
- sulfatase modifying factor 1
- transaldolase 1
- transmembrane anterior posterior transformation 1
- tubulin folding cofactor E
- T-box transcription factor 15
- T-box transcription factor 3
- T-box transcription factor 4
- T-box transcription factor 5
- T-box transcription factor 6
- thromboxane A synthase 1
- transcription factor 12
- T cell immune regulator 1, ATPase H⁺ transporting V0 subunit a3
- treacle ribosome biogenesis factor 1
- tectonic family member 2
- tectonic family member 3
- terminal nucleotidyltransferase 5A
- telomerase reverse transcriptase
- transforming growth factor beta 1
- transforming growth factor beta 2
- transforming growth factor beta receptor 1
- transforming growth factor beta receptor 2
- thrombopoietin
- transmembrane protein 165
- transmembrane protein 216
- transmembrane protein 231

- transmembrane protein 38B
- transmembrane protein 67
- TNF receptor superfamily member 11a
- TNF receptor superfamily member 11b
- TNF superfamily member 11
- tonsoku like, DNA repair protein
- tumor protein p63
- trafficking protein particle complex subunit 2
- triggering receptor expressed on myeloid cells 2
- thyroid hormone receptor interactor 11
- transcriptional repressor GATA binding 1
- transient receptor potential cation channel subfamily V member 4
- transient receptor potential cation channel subfamily V member 6
- tetratricopeptide repeat domain 21B
- tetratricopeptide repeat domain 8
- twist family bHLH transcription factor 1
- transmembrane immune signaling adaptor TYROBP
- vitamin D receptor
- VPS35 endosomal protein sorting factor like
- WD repeat containing planar cell polarity effector
- WD repeat domain 19
- WD repeat domain 35
- Wnt family member 1
- Wnt family member 10B
- Wnt family member 3
- Wnt family member 3A
- Wnt family member 5A
- Wnt family member 7A
- X-ray repair cross complementing 4
- xylosyltransferase 1
- xylosyltransferase 2
- zinc metalloproteinase STE24
- zinc finger SWIM-type containing 6

Related Gene Panels

- [Skeletal dysplasia - UGent](#)

Source URL: http://gentest.healthdata.be/genetic_test/1037