

GENETIC TEST: Skeletal dysplasia (gene panel)

FULL NAME:	Skeletal dysplasia (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal Diagnosis, Pre-implantation genetic diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Frozen tissue, Fresh tissue, Amniotic fluid, Chorionic villi, Cell culture, Skin fibroblasts
METHOD CATEGORY:	Sequence analysis: entire coding region Mutation screening and sequence analysis of selected exons

METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-10-07 / 2026-06-14
EQA:	• Skeletal dysplasia
TURNAROUND TIME (MAXIMUM):	6 months (2 months for prenatal testing)
CREATED:	28 Aug 2019 - 13:01
CHANGED:	09 Mar 2023 - 16:29
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Skelet dysplasie

Source URL: http://gentest.healthdata.be/genetic_test/784

RELATED CONTENT

Related Diseases

- [Isolated Klippel-Feil syndrome](#)

Related Laboratories

- [Centrum Medische Genetica - UZ Brussel VUB](#)

Related Analytes

- [ATP binding cassette subfamily C member 9](#)
- [ABL proto-oncogene 1, non-receptor tyrosine kinase](#)
- [aggrecan](#)
- [acid phosphatase 4](#)
- [acid phosphatase 5, tartrate resistant](#)
- [activin A receptor type 1](#)
- [ADAM metalloproteinase with thrombospondin type 1 motif 10](#)
- [ADAM metalloproteinase 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](#)
- [alkaline phosphatase, biomineralization associated](#)

- ALX homeobox 1
- ALX homeobox 3
- ALX homeobox 4
- APC membrane recruitment protein 1
- ANKH inorganic pyrophosphate transport regulator
- anoctamin 5
- ANTXR cell adhesion molecule 2
- APC regulator of WNT signaling pathway
- archain 1
- Rho GTPase activating protein 31
- AT-rich interaction domain 1A
- AT-rich interaction domain 1B
- arylsulfatase L
- activating signal cointegrator 1 complex subunit 1
- ASXL transcriptional regulator 2
- ATPase H⁺ transporting V0 subunit a2
- beta-1,3-galactosyltransferase 6
- beta-1,3-glucuronyltransferase 3
- beta-1,4-galactosyltransferase 7
- biglycan
- 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
- BRCA1 interacting helicase 1
- blood vessel epicardial substance
- complement C1r
- complement C1s
- 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 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
- 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
- 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
- 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
- centrosome and spindle pole associated protein 1
- cathepsin A
- cathepsin K
- cullin 7
- cytochrome P450 family 26 subfamily B member 1
- discoidin domain receptor tyrosine kinase 2
- DEAD-box helicase 59
- 24-dehydrocholesterol reductase
- dihydroorotate dehydrogenase (quinone)
- delta like canonical Notch ligand 3
- distal-less homeobox 3
- distal-less homeobox 5
- distal-less homeobox 6
- dentin matrix acidic phosphoprotein 1
- dedicator of cytokinesis 6
- dishevelled segment polarity protein 1
- dishevelled segment polarity protein 3
- dymeclin
- dynein cytoplasmic 2 heavy chain 1
- dynein 2 intermediate chain 2
- dynein cytoplasmic 2 light intermediate chain 1
- 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
- engulfment and cell motility 2
- 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
- enhancer of zeste 2 polycomb repressive complex 2 subunit
- FAM111 trypsin like peptidase A
- FAM20C golgi associated secretory pathway kinase
- FA complementation group A
- FA complementation group B
- FA complementation group C
- FA complementation group D2
- FA complementation group E
- FA complementation group F
- FA complementation group G
- FA complementation group I
- FA complementation group L
- 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
- fibronectin 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
- gap junction protein alpha 1
- galactosidase beta 1
- GLI family zinc finger 3
- GNAS complex locus
- glyceronephosphate O-acyltransferase
- 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 3
- glypican 6
- G protein-coupled receptor 68
- glutathione peroxidase 4
- gremlin 1, DAN family BMP antagonist
- glucuronidase beta
- histone deacetylase 4

- histone deacetylase 8
- hes family bHLH transcription factor 7
- hedgehog acyltransferase
- homeobox A11
- homeobox A13
- homeobox D13
- 15-hydroxyprostaglandin dehydrogenase
- heparan sulfate proteoglycan 2
- HECT, UBA and WWE domain containing E3 ubiquitin protein ligase 1
- isocitrate dehydrogenase (NADP(+)) 1
- isocitrate dehydrogenase (NADP(+)) 2
- iduronate 2-sulfatase
- interferon induced transmembrane protein 5
- intraflagellar transport 122
- intraflagellar transport 140
- intraflagellar transport 172
- intraflagellar transport 43
- intraflagellar transport 80
- insulin like growth factor 1 receptor
- 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
- jagged canonical Notch ligand 1
- lysine acetyltransferase 6B
- kinesin family member 22
- kinesin family member 7
- KRAS proto-oncogene, GTPase
- 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
- leucine rich repeat, Ig-like and transmembrane domains 3
- LDL receptor related protein 5
- latent transforming growth factor beta binding protein 2
- latent transforming growth factor beta binding protein 3
- mab-21 like 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
- matrilin 3
- multiple EGF like domains 8
- mesenchyme homeobox 1
- mesoderm posterior bHLH transcription factor 2
- matrix Gla protein
- melanocyte inducing transcription factor
- MKS transition zone complex subunit 1
- matrix metallopeptidase 13
- matrix metallopeptidase 2
- matrix metallopeptidase 9
- motor neuron and pancreas homeobox 1
- msh homeobox 2
- MYCN proto-oncogene, bHLH transcription factor
- myelin transcription factor 1
- N-acetyl-alpha-glucosaminidase
- 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 2
- NPR2 like, GATOR1 complex subunit
- nuclear receptor binding SET domain protein 1
- NAD(P) dependent steroid dehydrogenase-like
- neurotrophic receptor tyrosine kinase 2
- 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
- partner and localizer of BRCA2
- presequence translocase associated motor 16
- pantothenate kinase 2
- 3'-phosphoadenosine 5'-phosphosulfate synthase 2
- pericentrin
- phosphate cytidylyltransferase 1A, choline
- phosphodiesterase 3A
- phosphodiesterase 4D
- peroxisomal biogenesis factor 7
- phosphoglucomutase 3
- phosphate regulating endopeptidase X-linked
- phosphatidylinositol glycan anchor biosynthesis class V
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha
- paired like homeodomain 1
- pleckstrin homology and RUN domain containing M1
- procollagen-lysine,2-oxoglutarate 5-dioxygenase 2
- plastin 3

- 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
- prokineticin 2
- phosphatidylserine synthase 1
- parathyroid hormone 1 receptor
- parathyroid hormone like hormone
- protein tyrosine phosphatase non-receptor type 11
- pyrroline-5-carboxylate reductase 1
- RAB23, member RAS oncogene family
- RAB33B, member RAS oncogene family
- RAD21 cohesin complex component
- RAD51 paralog C
- RAS guanyl releasing protein 2
- RNA binding motif protein 8A
- recombination signal binding protein for immunoglobulin kappa J region
- RecQ like helicase 4
- RNA sensor RIG-I
- RNA component of mitochondrial RNA processing endoribonuclease
- RNA, U4atac small nuclear (U12-dependent splicing)
- receptor tyrosine kinase like orphan receptor 2
- RPGRIP1 like
- R-spondin 2
- ring finger and SPRY domain containing 1
- RUNX family transcription factor 2
- spalt like transcription factor 1
- spalt like transcription factor 4
- SBDS ribosome maturation factor
- 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
- SH3 domain binding protein 2
- SH3 and PX domains 2B
- SHOX homeobox
- 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 3
- solute carrier family 35 member D1
- solute carrier family 39 member 13
- solute carrier organic anion transporter family member 5A1
- secreted LY6/PLAUR domain containing 1
- SLX4 structure-specific endonuclease subunit
- SMAD family member 3
- SMAD family member 4
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 2
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 4
- 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 b, member 1
- structural maintenance of chromosomes 1A
- structural maintenance of chromosomes 3
- smoothened, frizzled class receptor
- small nuclear ribonucleoprotein polypeptides B and B1
- sorting nexin 10
- sclerostin
- SRY-box transcription factor 6
- SRY-box transcription factor 9
- Sp7 transcription factor
- signal transducer and activator of transcription 3
- sulfatase 1

- sulfatase modifying factor 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 3
- TDP-glucose 4,6-dehydratase
- transforming growth factor beta 1
- transforming growth factor beta 2
- transforming growth factor beta receptor 1
- transforming growth factor beta receptor 2
- thrombopoietin
- transmembrane and coiled-coil domains 1
- transmembrane protein 216
- transmembrane protein 38B
- transmembrane protein 67
- TNF receptor superfamily member 11b
- tumor protein p63
- trafficking protein particle complex subunit 2
- triggering receptor expressed on myeloid cells 2
- thyroid hormone receptor interactor 11
- thyroid hormone receptor interactor 4
- transcriptional repressor GATA binding 1
- transient receptor potential cation channel subfamily V member 4
- tetratricopeptide repeat domain 21B
- twist family bHLH transcription factor 1
- transmembrane immune signaling adaptor TYROBP

- [ubiquitin conjugating enzyme E2 T](#)
- [WD repeat domain 19](#)
- [WD repeat domain 35](#)
- [Wnt family member 1](#)
- [Wnt family member 10B](#)
- [Wnt family member 3](#)
- [Wnt family member 5A](#)
- [Wnt family member 6](#)
- [Wnt family member 7A](#)
- [xylosyltransferase 1](#)
- [xylosyltransferase 2](#)
- [zinc finger E-box binding homeobox 2](#)
- [Zic family member 1](#)
- [zinc metalloproteinase STE24](#)
- [zinc finger SWIM-type containing 6](#)

Related Gene Panels

- [Skeletal dysplasia \(394 genes\) - VUB](#)

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