Discover Dysplasias™
No-Charge Genetic Test for Skeletal Dysplasias

Discover Dysplasias Gene Panel

Discover Dysplasias tests 109 genes associated with skeletal dysplasias. This list is gathered from various sources, is not designed to be comprehensive, and is provided for reference only. This list is not medical advice and should not be used to make any diagnosis. Refer to lab reports in connection with potential diagnoses. Some genes below may also be associated with non-skeletal dysplasia disorders; those non-skeletal dysplasia disorders are not included on this list.

Skeletal Disorders Tested

Gene	Condition(s)	Inheritance
ACP5	Spondyloenchondrodysplasia with immune dysregulation (SED)	AR
ADAMTS10	Weill-Marchesani syndrome (WMS)	AR
AGPS	Rhizomelic chondrodysplasia punctata type 3 (RCDP)	AR
ALPL	Hypophosphatasia	AD/AR
ANKH	Craniometaphyseal dysplasia (CMD)	AD
ARSB	Mucopolysaccharidosis type VI (MPS VI), also known as Maroteaux-Lamy syndrome	AR
ARSE	Chondrodysplasia punctata	XLR
B3GALT6	Spondyloepimetaphyseal dysplasia with joint laxity type 1 (SEMDJL1) Ehlers-Danlos syndrome progeroid type 2 (EDSP2)	AR
B3GAT3	Multiple joint dislocations, short stature and craniofacial dysmorphism with or without congenital heart defects (JDSCD)	AR
BGN	Spondyloepimetaphyseal dysplasia (SEMD) Thoracic aortic aneurysm and dissection (TADD), with or without additional features, also known as Meester-Loeys syndrome	XL
BMP2	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies	AD
BMPR1B	Acromesomelic dysplasia Brachydactyly type A2 Brachydactyly type A1	AR AD AD
CANT1	Desbuquois dysplasia Multiple epiphyseal dysplasia (MED)	AR
CDC45	Meier-Gorlin syndrome	AR

CDC6	Meier-Gorlin syndrome, ***preliminary evidence*	AR
CDT1	Meier-Gorlin syndrome	AR
CFAP410	Axial spondylometaphyseal dysplasia (SMDAX)	AR
CHST14	CHST14-congenital disorder of glycosylation, also known as musculocontractural type Ehlers-Danlos syndrome or adducted thumb-clubfoot syndrome	AR
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations (SEDCJD)	AR
COL10A1	Metaphyseal chondrodysplasia, Schmid type (MCDS)	AD
COL11A1	Stickler syndrome Marshall syndrome Fibrochondrogenesis	AD/AR AD/AR AR
COL2A1	Achondrogenesis type II Stickler syndrome, type I Multiple forms of dysplasia, including Kniest dysplasia, Platyspondylic lethal skeletal dysplasia Torrance type, and Spondyloepimetaphyseal dysplasia Spondyloepiphyseal dysplasia congenita	AD AR
COL9A1	Stickler syndrome, type IV	AR
COL9A2	Stickler syndrome Multiple epiphyseal dysplasia (MED)	AR AD
COL9A3	Multiple epiphyseal dysplasia (MED) Stickler syndrome	AD AR
COMP	Multiple epiphyseal dysplasia (MED) Pseudoachondroplasia (PSACH)	AD
CUL7	3-M syndrome (3M)	AR
DDR2	Spondylometaepiphyseal dysplasia with short limbs and abnormal calcifications (SMED-SL)	AR
DLL3	Spondylocostal dysostosis	AR
DVL1	Robinow syndrome (ADRS)	AD
DVL3	Robinow syndrome (ADRS)	AD
DYM	Dyggve-Melchior-Clausen disease (DMC) Smith-McCort dysplasia	AR

EBP	Chondrodysplasia punctata type II (CDPX2) Male EBP disorder with neurological defects (MEND)	XLD XLR
EIF2AK3	Wolcott-Rallison syndrome (WRS)	AR
EVC	Ellis-van Creveld syndrome (EvC), a form of short-rib thoracic dysplasia	AR
EVC2	Ellis-van Creveld syndrome (EvC), a form of short-rib thoracic dysplasia Weyers acrodental dysostosis (WAD)	AR AD
FBN1	Marfan syndrome Weill-Marchesani syndrome Geleophysic dysplasia Acromicric dysplasia	AD
FGFR1	Craniosynostosis, including Pfeiffer syndrome Hartsfield syndrome Osteoglophonic dysplasia	AD
FGFR2	Apert syndrome Crouzon syndrome Jackson-Weiss syndrome Pfeiffer syndrome Beare-Stevenson syndrome Bent bone dysplasia	AD
FGFR3	Achondroplasia Camptodactyly, tall stature, and hearing loss (CATSHL) syndrome Crouzon syndrome with acanthosis nigricans Hypochondroplasia Muenke syndrome Severe achondroplasia with developmental delay and acanthosis nigricans (SADDAN) Thanatophoric dysplasia	AD
FLNA	Periventricular heterotopia with or without Ehlers-Danlos features Otopalatodigital spectrum disorders Frontometaphyseal dysplasia Melnick-Needles syndrome Terminal osseous dysplasia	XL
FLNB	Atelosteogenesis type I (AO1) Atelosteogenesis type III (AOIII) Boomerang dysplasia Piepkorn osteochondrodysplasia Larsen syndrome Spondylocarpotarsal synostosis syndrome (SCT)	AD AR

FN1	Spondylometaphyseal dysplasia - corner fracture type, also known as Sutcliffe type	AD
GALNS	Mucopolysaccharidosis type IVA (MPS IVA), also known as Morquio A	AR
GDF5	Brachydactyly and symphalangism Grebe syndrome Acromesomelic dysplasia, Hunter-Thompson type (AMDH) Du Pan syndrome	AD AR
GLB1	Mucopolysaccharidosis, type IVB (MPS IVB), also known as Morquio B	AR
GMNN	Meier-Gorlin syndrome	AD
GNPAT	Rhizomelic chondrodysplasia punctata type 2 (RCDP2)	AR
GNPTAB	Mucolipidosis type II alpha/beta (ML II) Mucolipidosis type III alpha/beta (ML III)	AR
GNPTG	Mucolipidosis type III gamma (ML III gamma)	AR
GNS	Mucopolysaccharidosis type IIID (MPS IIID), also known as Sanfilippo D	AR
GORAB	Geroderma osteodysplastica	AR
GPC6	Omodysplasia	AR
GUSB	Mucopolysaccharidosis type VII (MPS VII), also known as Sly syndrome	AR
HES7	Spondylocostal dysostosis	AR
HGSNAT	Mucopolysaccharidosis type IIIC (MPS IIIC), also known as Sanfilippo C	AR
HSPG2	Schwartz-Jampel syndrome type 1 (SJS1) Dyssegmental dysplasia, Silverman-Handmaker type (DDSH)	AR
HYAL1	Mucopolysaccharidosis type IX (MPS IX)	AR
ICK	Endocrine-cerebro-osteodysplasia (ECO)	AR
IDS	Mucopolysaccharidosis type II (MPS II), also known as Hunter syndrome	XLR
IDUA	Mucopolysaccharidosis type I (MPS I), including Hurler, Hurler-Scheie, and Scheie syndromes	AR
IFT172	Short-rib thoracic dysplasia 10 with or without polydactyly	AR
IHH	Brachydactyly type A1 (BDA1) Acrocapitofemoral dysplasia (ACFD)	AD AR

IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type	AR
INPPL1	Opsismodysplasia (OPSMD)	AR
KAT6B	Genitopatellar syndrome (GPS) Say-Barber-Biesecker-Young-Simpson syndrome (SBBYSS)	AD
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL)	AD
LBR	Greenberg dysplasia	AR
LEMD3	Buschke-Ollendorff syndrome (BOS) Osteopoikilosis, with or without melorheostosis	AD
LFNG	Spondylocostal dysostosis	AR
LIFR	Stuve-Wiedemann syndrome (SWS)	AR
LMX1B	Nail-patella syndrome (NPS)	AD
MAP3K7	Cardiospondylocarpofacial syndrome (CSCFS) Frontometaphyseal dysplasia (FMD)	AD
MATN3	Multiple epiphyseal dysplasia (MED) Spondyloepimetaphyseal dysplasia (SEMD), ** *preliminary evidence*	AD AR
MESP2	Spondylocostal dysostosis	AR
MGP	Keutel syndrome (KTLS)	AR
MMP13	Metaphyseal anadysplasia (MAD), including spondyloepimetaphyseal dysplasia, Missouri type (SEMD(MO)) Metaphyseal dysplasia, Spahr type (MDST)	AD/AR AR
MMP2	Multicentric osteolysis, nodulosis, and arthropathy (MONA)	AR
MMP9	Metaphyseal anadysplasia, ** *preliminary evidence*	AR
NAGLU	Mucopolysaccharidosis type IIIB (MPS IIIB), also known as Sanfilippo B	AR
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia (SMMD)	AR
NPR2	Acromesomelic dysplasia, Maroteaux type (AMDM) Epiphyseal chondrodysplasia, Miura type (ECDM)	AR AD
OBSL1	3-M syndrome (3M)	AR
ORC1	Meier-Gorlin syndrome	AR
ORC4	Meier-Gorlin syndrome	AR

ORC6	Meier-Gorlin syndrome	AR
PAPSS2	Brachyolmia (BCYM)	AR
PEX5	Zellweger spectrum disorder (ZSD) Rhizomelic chondrodysplasia punctata (RCDP)	AR
PEX7	Rhizomelic chondrodysplasia punctata (RCDP)	AR
PTH1R	Blomstrand chondrodysplasia (BOCD) Eiken syndrome Jansen type metaphyseal chondrodysplasia (JMC)	AR AD
RIPPLY2	Spondylocostal dysostosis	AR
RMRP	Cartilage-hair hypoplasia-anauxetic dysplasia (CHH-AD) spectrum disorders, including metaphyseal dysplasia without hypotrichosis	AR
ROR2	Brachydactyly type B1 (BDB1) Robinow syndrome	AD AR
RSPRY1	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type (SEMDFA)	AR
RUNX2	Cleidocranial dysplasia (CCD) Metaphyseal dysplasia with maxillary hypoplasia and brachydactyly	AD
SGSH	Mucopolysaccharidosis type IIIA (MPS IIIA), also known as Sanfilippo A	AR
SH3PXD2B	Frank-Ter Haar syndrome (FTHS)	AR
SLC26A2	Achondrogenesis, type IB (ACG1B) Atelosteogenesis type 2 (AO2) Diastrophic dysplasia (DTD) Multiple epiphyseal dysplasia 4 (EDM4)	AR
SLC39A13	Ehlers-Danlos syndrome-like spondylocheirodysplasia (SCD-EDS)	AR
SMAD4	Myhre syndrome	AD
SMARCAL1	Schimke immunoosseous dysplasia (SIOD)	AR
SOX9	Campomelic dysplasia	AD
TBCE	Kenney-Caffey syndrome (KCS)	AR
TBX6	Spondylocostal dysostosis, ** *preliminary evidence*	AD
TRPV4	Skeletal dysplasias, including Spondylometaphyseal dysplasia, Kozlowski type	AD

WDR35	Short-rib thoracic dysplasia (SRTD) with or without polydactyly, including cranioectodermal dysplasia	AR
WISP3	Progressive pseudorheumatoid dysplasia (PPRD)	AR
WNT5A	Robinow syndrome type (ADRS)	AD
XYLT1	Desbuquois dysplasia type 2	AR

Disorders Tested Table: AD (autosomal dominant), AR (autosomal recessive), XLD (X-linked dominant), XLR (X-linked recessive), **preliminary evidence (Preliminary-evidence genes currently have early evidence of a clinical association with the specific disease covered by this test.)

Inheritance

Skeletal dysplasias can occur in several inheritance patterns including autosomal dominant, autosomal recessive and X-linked.

Autosomal dominant (AD) disorders are caused by genes located on one of the numbered, or non-sex, chromosomes. Individuals with a disease-causing variant in one copy of the gene will be affected with or at risk for developing the disorder.

Autosomal recessive (AR) disorders are caused by genes located on one of the numbered, or non-sex, chromosomes. In order for an individual to be affected with an autosomal recessive disorder, they must have two disease-causing variants, one in each copy of the gene. Carriers of the disorder, who have only one disease-causing variant, typically do not have symptoms.

X-linked disorders are caused by genes located on the X chromosome. Females have two X chromosomes, while males have one X and one Y chromosome.

X-linked dominant (XLD) is a mode of inheritance in which individuals with a disease-causing variant in one copy of the X-linked gene will be affected with or at risk for developing the disorder.

X-linked recessive (XLR) is a mode of inheritance in which males with a single disease-causing variant are affected because the variant impacts their only copy of the gene. Females with two disease-causing variants, one in each copy of the gene, will also be affected. Carrier females typically do not have symptoms because they have a working copy of the gene on their other X chromosome; however, skewed X-inactivation can lead to varying degrees of clinical expression in carrier females.

Considerations for testing

This test may be appropriate for individuals with clinical findings suggestive of or consistent with a diagnosis of skeletal dysplasias. These clinical findings may include short stature, disproportionate growth, dysmorphic facial features and skeletal abnormalities.

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