Skeletal dysplasia

Gene panel

Gene panel information

Gene panel	**Skeletal dysplasia**
Version	5
Total genes	663
Activation date	Wednesday 28 october 2026
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABCC9	99.79 %	601439	Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive
ABL1	99.94 %	189980	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation; Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant
ACAN	91.04 %	155760	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3), Autosomal dominant; Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 (3), Autosomal recessive
ACP5	99.91 %	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACTB	99.98 %	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; Becker nevus, syndromic or isolated, somatic mosaic, 604919 (3); Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 (3), Autosomal dominant; Dystonia-deafness syndrome 1, 607371 (3), Autosomal dominant; Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470 (3)
ACTG1	100 %	102560	Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant
ACVR1	99.67 %	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ADAMTS10	99.84 %	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
ADAMTS17	99.86 %	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
ADAMTSL2	99.69 %	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
AFF3	99.09 %	601464	KINSSHIP syndrome, 619297 (3), Autosomal dominant
AFF4	99.87 %	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
AGA	99.77 %	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGPS	98.17 %	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
AHDC1	99.99 %	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
AIFM1	99.79 %	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive

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AKT1	99.96 %	164730	Proteus syndrome, somatic mosaic, 176920 (3); Breast cancer, somatic, 114480 (3); Cowden syndrome 6, 615109 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Ovarian cancer, somatic, 167000 (3)
AKT2	99.9 %	164731	Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant
ALG12	99.92 %	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
ALG3	99.96 %	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
ALG9	99.52 %	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive
ALPL	99.53 %	171760	Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant
ALX1	98.39 %	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
ALX3	98.56 %	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
ALX4	99.89 %	605420	Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive
AMER1	99.95 %	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
ANAPC1	72.56 %	608473	Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive
ANKH	99.76 %	605145	Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant
ANKRD11	99.54 %	611192	KBG syndrome, 148050 (3), Autosomal dominant
ANO5	99.62 %	608662	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant
ANTXR2	99.56 %	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
APC2	99.7 %	612034	Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 74, 617169 (3), Autosomal recessive
ARCN1	99.9 %	600820	Short stature-micrognathia syndrome, 617164 (3), Autosomal dominant
ARHGAP25	99.96 %	610587	*No OMIM phenotypes*
ARHGAP31	99.98 %	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARID1A	99.61 %	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	99.8 %	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARL6	99.89 %	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
ARSB	99.79 %	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
ARSK	99.74 %	610011	Mucopolysaccharidosis, type X, 619698 (3), Autosomal recessive
ARSL	99.68 %	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ASAH1	99.6 %	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
ASXL1	99.93 %	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
ASXL2	99.83 %	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
ATP6V0A2	99.77 %	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive

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ATP7A	99.79 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
ATR	99.75 %	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant
B3GALT6	99.98 %	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3), Autosomal recessive
B3GAT3	99.42 %	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
B3GLCT	99.97 %	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
B4GALT7	99.6 %	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
B9D1	99.49 %	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
B9D2	99.25 %	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
BANF1	97.05 %	603811	Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive
BBS1	99.96 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	99.8 %	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	99.98 %	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	99.4 %	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
BBS4	99.38 %	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	98.81 %	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	99.54 %	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	99.75 %	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCL11B	99.95 %	606558	Immunodeficiency 49, severe combined, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant
BGN	99.87 %	301870	Meester-Loeys syndrome, 300989 (3), X-linked; Spondyloepimetaphyseal dysplasia, X-linked, 300106 (3), X-linked recessive
BHLHA9	99.36 %	615416	?Camptosynpolydactyly, complex, 607539 (3), Autosomal recessive; Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3), Autosomal recessive
BMP1	99.61 %	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
BMP2	99.76 %	112261	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive
BMP4	99.99 %	112262	Orofacial cleft 11, 600625 (3), Autosomal dominant; Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
BMPER	99.68 %	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
BMPR1B	99.82 %	603248	Acromesomelic dysplasia 3, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant
BNIP1	99.84 %	603291	Spondylopeiphyseal dysplasia, Holling type, 621345 (3), Autosomal recessive

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BPNT2	99.96 %	614010	Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive
BRAF	99.53 %	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
C2CD3	99.73 %	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
CA2	97.98 %	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CANT1	99.62 %	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive
CASR	99.93 %	601199	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant; {?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3), Autosomal dominant
CBFB	99.83 %	121360	Cleidocranial dysplasia 2, 620099 (3), Autosomal dominant
CC2D2A	99.85 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCDC134	99.99 %	618788	Osteogenesis imperfecta, type XXII, 619795 (3), Autosomal recessive
CCDC8	99.89 %	614145	3-M syndrome 3, 614205 (3), Autosomal recessive
CCN6	99.9 %	603400	Progressive pseudorheumatoid dysplasia, 208230 (3), Autosomal recessive
CCNQ	99.66 %	300708	STAR syndrome, 300707 (3), X-linked dominant
CD96	99.72 %	606037	C syndrome, 211750 (3), Autosomal dominant
CDC45	98.79 %	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
CDC6	99.45 %	602627	Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
CDC73	98.96 %	607393	Hyperparathyroidism, familial primary, 145000 (3), Autosomal dominant; Parathyroid adenoma with cystic changes, 145001 (3), Autosomal dominant; Parathyroid carcinoma, 608266 (3); Hyperparathyroidism-jaw tumor syndrome, 145001 (3), Autosomal dominant
CDH3	99.86 %	114021	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive
CDK10	99.58 %	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
CDKN1C	99.99 %	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CDT1	99.68 %	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
CENPE	99.82 %	117143	?Microcephaly 13, primary, autosomal recessive, 616051 (3), Autosomal recessive
CEP120	99.85 %	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
CEP152	99.94 %	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
CEP290	98.33 %	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive

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CFAP410	99.95 %	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
CHD7	99.9 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHST11	99.99 %	610128	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167 (3), Autosomal recessive
CHST14	99.87 %	608429	Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive
CHST3	99.92 %	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive
CHSY1	99.99 %	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
CILK1	99.45 %	612325	Cranioectodermal dysplasia 6, 621337 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive
CKAP2L	99.44 %	616174	Filippi syndrome, 272440 (3), Autosomal recessive
CLCN5	99.79 %	300008	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Dent disease 1, 300009 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive
CLCN7	99.83 %	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant
COG1	99.95 %	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
COG4	99.68 %	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant
COL10A1	99.94 %	120110	Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant
COL11A1	97.48 %	120280	Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; Stickler syndrome, type II, 604841 (3), Autosomal dominant; Marshall syndrome, 154780 (3), Autosomal dominant; Deafness, autosomal dominant 37, 618533 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3)
COL11A2	99.65 %	120290	Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal recessive, Autosomal dominant; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant
COL1A1	99.22 %	120150	Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; {Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant

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COL1A2	98.99 %	120160	Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant
COL27A1	99.6 %	608461	Steel syndrome, 615155 (3), Autosomal recessive
COL2A1	99.31 %	120140	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Algerian type, 184253 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant
COL9A1	99.84 %	120210	Stickler syndrome, type IV, 614134 (3), Autosomal recessive; ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant
COL9A2	98.61 %	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
COL9A3	99.7 %	120270	{Intervertebral disc disease, susceptibility to}, 603932 (3); Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; Stickler syndrome, type VI, 620022 (3), Autosomal recessive
COLEC10	99.96 %	607620	3MC syndrome 3, 248340 (3), Autosomal recessive
COLEC11	99.85 %	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
COMP	99.77 %	600310	Pseudoachondroplasia, 177170 (3), Autosomal dominant; Carpal tunnel syndrome 2, 619161 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant
COPB2	99.58 %	606990	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 (3), Autosomal dominant; ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive
CPLANE1	99.74 %	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive
CREB3L1	99.3 %	616215	Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive
CREBBP	99.79 %	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRIPT	99.97 %	604594	Rothmund-Thomson syndrome, type 3, 615789 (3), Autosomal recessive
CRTAP	99.84 %	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
CSF1R	99.71 %	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant
CSGALNACT1	99.95 %	616615	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870 (3), Autosomal recessive

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CSPP1	99.85 %	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CTSA	99.77 %	613111	Galactosialidosis, 256540 (3), Autosomal recessive; Brain small vessel disease 6 with leukoencephalopathy, 621394 (3), Autosomal dominant
CTSC	99.79 %	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive
CTSK	99.31 %	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
CUL7	99.89 %	609577	3-M syndrome 1, 273750 (3), Autosomal recessive
CYP26B1	99.92 %	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (3)
CYP27B1	99.52 %	609506	Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive
CYP2R1	99.94 %	608713	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081 (3), Autosomal recessive
CYP3A4	99.44 %	124010	Vitamin D-dependent rickets, type 3, 619073 (3), Autosomal dominant
DCC	99.9 %	120470	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive
DDR1	99.82 %	600408	*No OMIM phenotypes*
DDR2	98.65 %	191311	Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant; Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive
DDRGK1	99.9 %	616177	Spondyloepimetaphyseal dysplasia, Shohat type, 602557 (3), Autosomal recessive
DDX58	99.64 %	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
DDX59	99.38 %	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DHCR24	99.28 %	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	99.89 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHODH	99.75 %	126064	Miller syndrome, 263750 (3), Autosomal recessive
DHRS3	99.8 %	612830	*No OMIM phenotypes*
DIP2C	99.9 %	611380	*No OMIM phenotypes*
DIS3L2	99.75 %	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DLL3	99.67 %	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive
DLL4	99.71 %	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DLX3	99.28 %	600525	Trichodontoosseous syndrome, 190320 (3), Autosomal dominant; Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant
DLX5	99.09 %	600028	Split-hand/foot malformation 1, 183600 (3), Autosomal dominant; ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
DLX6	99.8 %	600030	*No OMIM phenotypes*
DMP1	100 %	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
DNA2	99.6 %	601810	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 (3), Autosomal dominant; Rothmund-Thomson syndrome, type 4, 620819 (3), Autosomal recessive; Seckel syndrome 8, 615807 (3), Autosomal recessive
DNAJC21	99.71 %	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
DNMT3A	99.69 %	602769	Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DOCK6	99.4 %	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DONSON	99.84 %	611428	Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive
DPAGT1	99.95 %	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive
DPM1	96.61 %	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DSE	99.99 %	605942	Ehlers-Danlos syndrome, musculocontractural type 2, 615539 (3), Autosomal recessive
DSPP	99.98 %	125485	Dentinogenesis imperfecta, Shields type III, 125500 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type II, 125490 (3), Autosomal dominant; Dentin dysplasia, type II, 125420 (3), Autosomal dominant; Deafness, autosomal dominant 39, with dentinogenesis, 605594 (3), Autosomal dominant
DVL1	99.85 %	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
DVL3	99.83 %	601368	Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant
DYM	99.76 %	607461	Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive
DYNC2H1	99.74 %	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
DYNC2I1	99.94 %	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
DYNC2I2	99.68 %	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
DYNC2LI1	99.86 %	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
DYNLT2B	99.98 %	617353	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive
EBP	99.7 %	300205	MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant
EDN1	99.89 %	131240	Question mark ears, isolated, 612798 (3), Autosomal dominant; Auriculocondylar syndrome 3, 615706 (3), Autosomal recessive
EDNRA	99.92 %	131243	{Migraine, resistance to}, 157300 (3), Autosomal dominant; Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant
EDNRB	99.98 %	131244	{Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; ?ABCD syndrome, 600501 (3), Autosomal recessive; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant
EED	99.59 %	605984	Cohen-Gibson syndrome, 617561 (3), Autosomal dominant
EFL1	98.86 %	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
EFNA4	97.83 %	601380	*No OMIM phenotypes*
EFNB1	99.61 %	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
EFTUD2	99.55 %	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EIF2AK3	98.74 %	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
EIF4A3	99.96 %	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
ELMO2	99.98 %	606421	Vascular malformation, primary intraosseous, 606893 (3), Autosomal recessive
EN1	99.98 %	131290	?ENDOVE syndrome, limb-brain type, 619218 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ENPP1	99.81 %	173335	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant
EOGT	98.82 %	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
EP300	99.9 %	602700	Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EPS15L1	99.83 %	616826	*No OMIM phenotypes*
ERCC4	99.73 %	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive
ERF	99.68 %	611888	Craniosynostosis 4, 600775 (3), Autosomal dominant; Chitayat syndrome, 617180 (3), Autosomal dominant
ESCO2	99.95 %	609353	Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive
EVC	99.73 %	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	99.79 %	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EXOC6B	99.39 %	607880	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395 (3), Autosomal recessive
EXT1	99.88 %	608177	Exostoses, multiple, type 1, 133700 (3), Autosomal dominant; Chondrosarcoma, 215300 (3), Somatic mutation
EXT2	99.84 %	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
EXTL3	99.94 %	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
EZH2	99.77 %	601573	Weaver syndrome, 277590 (3), Autosomal dominant
FAM111A	99.98 %	615292	Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant
FAM20B	99.77 %	611063	*No OMIM phenotypes*
FAM20C	99.92 %	611061	Raine syndrome, 259775 (3), Autosomal recessive
FANCB	99.51 %	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
FAR1	99.8 %	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
FBLN1	99.78 %	135820	*No OMIM phenotypes*
FBN1	99.73 %	134797	Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant
FBN2	99.55 %	612570	Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant
FBXW4	99.5 %	608071	*No OMIM phenotypes*
FERMT3	99.73 %	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FGD1	99.65 %	300546	Aarskog-Scott syndrome, 305400 (3), X-linked recessive
FGF10	99.83 %	602115	LADD syndrome 3, 620193 (3), Autosomal dominant; Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant
FGF16	99.54 %	300827	Metacarpal 4-5 fusion, 309630 (3), X-linked recessive
FGF23	99.92 %	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3), Autosomal recessive; Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant
FGF3	99.74 %	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
FGF4	99.45 %	164980	Short-rib thoracic dysplasia 22 without polydactyly, 621260 (3), Autosomal recessive
FGF8	99.65 %	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGF9	99.96 %	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant
FGFR1	99.77 %	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FGFR2	99.76 %	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant
FGFR3	99.87 %	134934	Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); LADD syndrome 2, 620192 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant
FIG4	99.67 %	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive
FKBP10	99.7 %	607063	Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive; Bruck syndrome 1, 259450 (3), Autosomal recessive
FKBP14	99.96 %	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive

Skeletal dysplasia

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FLNA	99.9 %	300017	Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
FLNB	99.84 %	603381	Larsen syndrome, 150250 (3), Autosomal dominant; Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive; Boomerang dysplasia, 112310 (3), Autosomal dominant
FMN1	99.97 %	136535	*No OMIM phenotypes*
FN1	99.65 %	135600	Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant; Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant
FOSL2	99.72 %	601575	Aplasia cutis-enamel dysplasia syndrome, 620789 (3), Autosomal dominant
FOXC1	99.97 %	601090	Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant
FREM1	99.89 %	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Bifid nose with or without anorectal and renal anomalies, 608980 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant
FUCA1	98.82 %	612280	Fucosidosis, 230000 (3), Autosomal recessive
FUZ	99.56 %	610622	{Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant
FZD2	99.89 %	600667	Omodysplasia 2, 164745 (3), Autosomal dominant
GALNS	99.66 %	612222	Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive
GALNT3	99.16 %	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
GBA	93.64 %	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
GCM2	99.96 %	603716	Hypoparathyroidism, familial isolated 2, 618883 (3), Autosomal recessive, Autosomal dominant; Hyperparathyroidism 4, 617343 (3), Autosomal dominant
GDF3	99.99 %	606522	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia, isolated, with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant
GDF5	99.97 %	601146	Acromesomelic dysplasia 2A, 200700 (3), Autosomal recessive; Acromesomelic dysplasia 2B, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; Symphalangism, proximal, 1B, 615298 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; ?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type C, 113100 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GDF6	99.99 %	601147	Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant
GFER	99.98 %	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive
GHR	99.52 %	600946	Laron dwarfism, 262500 (3), Autosomal recessive; Increased responsiveness to growth hormone, 604271 (3), Autosomal dominant; Growth hormone insensitivity, partial, 604271 (3), Autosomal dominant; {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal recessive, Autosomal dominant
GINS2	99.77 %	610609	*No OMIM phenotypes*
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GLB1	99.98 %	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLI1	99.29 %	165220	Polydactyly, preaxial I, 174400 (3), Autosomal recessive; Polydactyly, postaxial, type A8, 618123 (3), Autosomal recessive
GLI2	99.67 %	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
GLI3	99.97 %	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GMNN	99.29 %	602842	Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant
GNAI3	98.72 %	139370	Auriculocondylar syndrome 1, 602483 (3), Autosomal dominant
GNAS	99.97 %	139320	Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; McCune-Albright syndrome, somatic mosaic, 174800 (3); Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant; ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080 (3)
GNPAT	99.73 %	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPNAT1	100 %	616510	?Rhizomelic dysplasia, Ain-Naz type, 619598 (3), Autosomal recessive
GNPTAB	99.54 %	607840	Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive
GNPTG	99.93 %	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GNS	98.91 %	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GORAB	99.51 %	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive
GPC3	99.69 %	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
GPC4	97.57 %	300168	Keipert syndrome, 301026 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GPC6	99.73 %	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
GPX4	99.94 %	138322	Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive
GREM1	99.99 %	603054	*No OMIM phenotypes*
GRK2	99.75 %	109635	*No OMIM phenotypes*
GSC	99.86 %	138890	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 (3), Autosomal recessive
GUSB	99.73 %	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
GZF1	99.98 %	613842	Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive
HAAO	99.28 %	604521	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive
HDAC4	99.89 %	605314	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797 (3), Autosomal dominant
HDAC8	99.57 %	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HES7	99.74 %	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
HGSNAT	99.82 %	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
HHAT	99.88 %	605743	Nivelon-Nivelon-Mabille syndrome, 600092 (3), Autosomal recessive
HNRNPC	99.81 %	164020	Intellectual developmental disorder, autosomal dominant 74, 620688 (3), Autosomal dominant
HNRNPK	99.59 %	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
HOXA11	99.71 %	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant
HOXA13	99.85 %	142959	Hand-foot-genital syndrome, 140000 (3), Autosomal dominant; ?Guttmacher syndrome, 176305 (3), Autosomal dominant
HOXD13	99.85 %	142989	Syndactyly, type V, 186300 (3), Autosomal dominant; Synpolydactyly 1, 186000 (3), Autosomal dominant; Brachydactyly, type E, 113300 (3), Autosomal dominant; Brachydactyly, type D, 113200 (3), Autosomal dominant; ?Brachydactyly-syndactyly syndrome, 610713 (3)
HPGD	99.85 %	601688	?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive
HS2ST1	93.17 %	604844	Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive
HSPA9	99.8 %	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
HSPG2	99.18 %	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
HUWE1	99.73 %	300697	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked
HYAL1	99.88 %	607071	Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive
IARS2	99.59 %	612801	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
ID4	98.89 %	600581	*No OMIM phenotypes*
IDH1	99.85 %	147700	{Glioma, susceptibility to, somatic}, 137800 (3)
IDH2	99.95 %	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDS	99.17 %	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IDUA	99.91 %	252800	Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive
IFIH1	99.64 %	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFITM5	99.96 %	614757	Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant
IFT122	99.49 %	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	99.77 %	614620	{Polycystic kidney disease 9, susceptibility to}, 621164 (3), Autosomal dominant; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Cranioectodermal dysplasia 5, 621180 (3), Autosomal recessive
IFT172	99.83 %	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT43	99.83 %	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive
IFT52	99.81 %	617094	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive
IFT80	99.71 %	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IFT81	94.88 %	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
IHH	99.91 %	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant
IKBKG	54.61 %	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked
IL11RA	99.83 %	600939	Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive
IL1RN	99.11 %	147679	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852 (3), Autosomal recessive; {Gastric cancer risk after H. pylori infection}, 613659 (3); {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
IL6ST	99.8 %	600694	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752 (3), Autosomal dominant; Stuve-Wiedemann syndrome 2, 619751 (3), Autosomal recessive; Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523 (3), Autosomal recessive; ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 (3), Autosomal dominant
INPPL1	99.35 %	600829	Opsismodysplasia, 258480 (3), Autosomal recessive
INTU	99.87 %	610621	?Orofaciodigital syndrome XVII, 617926 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 20 with polydactyly, 617925 (3), Autosomal recessive
JAG1	99.94 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KAT6B	99.82 %	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KCNJ2	100 %	600681	Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Andersen syndrome, 170390 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3), Autosomal dominant
KDELR2	99.83 %	609024	Osteogenesis imperfecta, type XXI, 619131 (3), Autosomal recessive
KDM6A	99.7 %	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KIAA0586	99.33 %	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
KIAA0753	99.95 %	617112	?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive; ?Joubert syndrome 38, 619476 (3), Autosomal recessive; Short-rib thoracic dysplasia 21 without polydactyly, 619479 (3), Autosomal recessive
KIAA0825	99.85 %	617266	Polydactyly, postaxial, type A10, 618498 (3), Autosomal recessive
KIAA1217	99.95 %	617367	*No OMIM phenotypes*
KIF22	99.68 %	603213	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3), Autosomal dominant
KIF24	99.87 %	613747	*No OMIM phenotypes*
KIF5B	99.82 %	602809	*No OMIM phenotypes*
KIF7	99.84 %	611254	Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
KL	99.89 %	604824	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 (3), Autosomal recessive
KMT2D	99.72 %	602113	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant
KRAS	97.26 %	190070	Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)
KYNU	99.13 %	605197	?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive
LAMA5	99.73 %	601033	Nephrotic syndrome, type 26, 620049 (3), Autosomal recessive; ?Bent bone dysplasia syndrome 2, 620076 (3), Autosomal recessive
LBR	99.84 %	600024	Pelger-Huet anomaly, 169400 (3), Autosomal dominant; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 (3), Autosomal recessive; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive
LEMD3	98.11 %	607844	Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant; Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant
LFNG	99.97 %	602576	Spondylocostal dysostosis 3, autosomal recessive, 609813 (3), Autosomal recessive
LIFR	99.8 %	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
LMBR1	99.88 %	605522	Syndactyly, type IV, 186200 (3), Autosomal dominant; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Acheiropody, 200500 (3), Autosomal recessive; Triphalangeal thumb-polysyndactyly syndrome, 190605 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LMNA	99.84 %	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
LMOD3	99.97 %	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
LMX1B	99.92 %	602575	Focal segmental glomerulosclerosis 10, 256020 (3), Autosomal dominant; Nail-patella syndrome, 161200 (3), Autosomal dominant
LONP1	99.79 %	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LPIN2	99.9 %	605519	Majeed syndrome, 609628 (3)
LRP1	99.42 %	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive; Developmental dysplasia of the hip 3, 620690 (3), Autosomal dominant
LRP4	99.58 %	604270	?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive
LRP5	99.56 %	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Endosteal hyperostosis, 144750 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; [Bone mineral density variability 1, high bone mass], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant
LRRK1	99.93 %	610986	Osteosclerotic metaphyseal dysplasia, 615198 (3), Autosomal recessive
LTBP1	99.63 %	150390	Cutis laxa, autosomal recessive, type IIE, 619451 (3), Autosomal recessive
LTBP2	99.69 %	602091	Glaucoma 3, primary congenital, D, 613086 (3); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive
LTBP3	99.41 %	602090	Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant
MAB21L2	99.99 %	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant
MAFB	99.57 %	608968	Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant
MAN2B1	99.51 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAN2B2	99.82 %	618899	Congenital disorder of glycosylation type 1EE with or without immunodeficiency, 621140 (3), Autosomal recessive
MAN2C1	99.64 %	154580	Congenital disorder of deglycosylation 2, 619775 (3), Autosomal recessive
MANBA	99.66 %	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
MAP2K1	99.43 %	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP3K20	99.66 %	609479	Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MAP3K7	98.99 %	602614	Frontometaphyseal dysplasia 2, 617137 (3), Autosomal dominant; Cardiospondylocarpofacial syndrome, 157800 (3), Autosomal dominant
MAPKAPK5	99.78 %	606723	Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive
MASP1	99.57 %	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
MATN3	99.65 %	602109	{Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant
MBTPS1	99.4 %	603355	Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392 (3), Autosomal recessive; CAOP syndrome, 621252 (3), Autosomal recessive
MBTPS2	99.85 %	300294	Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
MCM3	99.82 %	602693	*No OMIM phenotypes*
MCM5	99.8 %	602696	?Meier-Gorlin syndrome 8, 617564 (3), Autosomal recessive
MCM7	99.5 %	600592	*No OMIM phenotypes*
MECOM	99.85 %	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant
MEGF8	99.36 %	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEOX1	99.87 %	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
MESD	99.71 %	607783	Osteogenesis imperfecta, type XX, 618644 (3), Autosomal recessive
MESP2	99.91 %	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive
MET	99.84 %	164860	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3); ?Arthrogryposis, distal, type 11, 620019 (3), Autosomal dominant; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); {Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; ?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive
MGP	99.98 %	154870	Keutel syndrome, 245150 (3), Autosomal recessive
MIA2	93.37 %	602132	*No OMIM phenotypes*
MIA3	99.84 %	613455	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269 (3), Autosomal recessive
MID1	99.87 %	300552	Opitz GBBB syndrome, 300000 (3), X-linked recessive
MIR140	99.72 %	611894	Spondyloepiphyseal dysplasia, Nishimura type, 618618 (3), Autosomal dominant
MIR17HG	0 %	609415	*No OMIM phenotypes*
MKKS	99.99 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKS1	99.62 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MMP13	99.5 %	600108	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant; Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive
MMP14	99.79 %	600754	Winchester syndrome, 277950 (3), Autosomal recessive
MMP2	99.16 %	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
MMP9	99.8 %	120361	Metaphyseal anadysplasia 2, 613073 (3), Autosomal recessive
MNX1	99.01 %	142994	Currarino syndrome, 176450 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MPDU1	99.91 %	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MSX2	99.96 %	123101	Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant; Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina 1, 168500 (3), Autosomal dominant
MTAP	99.59 %	156540	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250 (3), Autosomal dominant
MTX2	99.38 %	608555	Mandibuloacral dysplasia progeroid syndrome, 619127 (3), Autosomal recessive
MYCN	99.94 %	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant; Megalencephaly-polydactyly syndrome, 620748 (3), Autosomal dominant
MYH3	99.96 %	160720	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant; Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 (3), Autosomal recessive; Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 (3), Autosomal dominant; Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 (3), Autosomal dominant
MYLPF	98.49 %	617378	Arthrogryposis, distal, type 1C, 619110 (3), Autosomal recessive, Autosomal dominant
MYO18B	99.89 %	607295	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive
NADSYN1	99.53 %	608285	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845 (3), Autosomal recessive
NAGLU	99.86 %	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
NANS	99.82 %	605202	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 (3), Autosomal recessive
NBAS	99.73 %	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
NEK1	99.87 %	604588	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive; ?Orofaciodigital syndrome II, 252100 (3), Autosomal recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant
NEK9	99.84 %	609798	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 (3), Autosomal recessive; Nevus comedonicus, somatic, 617025 (3); Lethal congenital contracture syndrome 10, 617022 (3), Autosomal recessive
NEPRO	99.47 %	617089	Anauxetic dysplasia 3, 618853 (3), Autosomal recessive
NEU1	99.94 %	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
NF1	99.83 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
NFIA	97.78 %	600727	Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant
NFIX	99.82 %	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant
NIPBL	99.72 %	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKX3-2	99.62 %	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NLRP3	99.95 %	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NMNAT1	99.58 %	608700	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 (3), Autosomal recessive; Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
NOG	99.02 %	602991	Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Brachydactyly, type B2, 611377 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant
NOTCH1	99.8 %	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NOTCH2	98.36 %	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NPPC	99.86 %	600296	*No OMIM phenotypes*
NPR2	99.89 %	108961	Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant; Acromesomelic dysplasia 1, Maroteaux type, 602875 (3), Autosomal recessive
NPR3	99.96 %	108962	Boudin-Mortier syndrome, 619543 (3), Autosomal recessive
NRAS	99.69 %	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NSD1	99.93 %	606681	Sotos syndrome, 117550 (3), Autosomal dominant
NSDHL	99.66 %	300275	CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant
NSMCE2	99.93 %	617246	Seckel syndrome 10, 617253 (3), Autosomal recessive
NUF2	96.39 %	611772	*No OMIM phenotypes*
NXN	99.94 %	612895	Robinow syndrome, autosomal recessive 2, 618529 (3), Autosomal recessive
OBSL1	99.88 %	610991	3-M syndrome 2, 612921 (3), Autosomal recessive
OFD1	99.71 %	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive
ORC1	98.12 %	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
ORC4	99.59 %	603056	Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive
ORC6	99.1 %	607213	Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive
OSTM1	99.58 %	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
P3H1	99.36 %	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
P4HB	99.98 %	176790	Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant
PAM16	99.81 %	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive
PAN2	99.43 %	617447	Developmental delay with variable cardiac and renal congenital anomalies and dysmorphic facies, 621384 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PAPSS2	99.86 %	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
PAX3	99.62 %	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
PCNT	99.91 %	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PCYT1A	99.93 %	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 5, 620680 (3), Autosomal recessive
PDE3A	99.9 %	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
PDE4D	99.76 %	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
PDGFRB	99.81 %	173410	Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; ?Ocular pterygium-digital keloid dysplasia syndrome, 621091 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant
PEX5	99.93 %	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
PEX7	99.68 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PFN1	99.99 %	176610	Amyotrophic lateral sclerosis 18, 614808 (3)
PGM3	99.78 %	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHEX	99.73 %	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
PHGDH	99.34 %	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHLDB1	99.88 %	612834	Osteogenesis imperfecta, type XXIII, 620639 (3), Autosomal recessive
PIGT	99.83 %	610272	?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Somatic mutation, Autosomal dominant; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive
PIGV	99.98 %	610274	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 (3), Autosomal recessive
PIK3C2A	99.81 %	603601	Oculoskeletodental syndrome, 618440 (3), Autosomal recessive
PIK3CA	99.63 %	171834	Hemifacial myohyperplasia, somatic, 606773 (3); CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Cerebral cavernous malformations 4, somatic, 619538 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Nevus, epidermal, somatic mosaic, 162900 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3)
PIK3R1	99.65 %	171833	Immunodeficiency 36, 616005 (3), Autosomal dominant; SHORT syndrome, 269880 (3), Autosomal dominant; Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive
PISD	99.94 %	612770	Liberfarb syndrome, 618889 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PITX1	99.83 %	602149	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 (3), Autosomal dominant
PKDCC	99.25 %	614150	Rhizomelic limb shortening with dysmorphic features, 618821 (3), Autosomal recessive
PLCB3	99.82 %	600230	Spondylometaphyseal dysplasia with corneal dystrophy, 618961 (3), Autosomal recessive
PLCB4	99.95 %	600810	Auriculocondylar syndrome 2B, 620458 (3), Autosomal recessive; Auriculocondylar syndrome 2A, 614669 (3), Autosomal dominant
PLEKHM1	99.02 %	611466	?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant
PLOD1	98.95 %	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive
PLOD2	99.59 %	601865	Bruck syndrome 2, 609220 (3), Autosomal recessive
PLS3	99.74 %	300131	Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant; Diaphragmatic hernia 5, X-linked, 306950 (3), X-linked
POC1A	99.49 %	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POLE	99.91 %	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive
POLR1A	99.5 %	616404	Leukodystrophy, hypomyelinating, 27, 620675 (3), Autosomal recessive; Acrofacial dysostosis, Cincinnati type, 616462 (3), Autosomal dominant
POLR1B	99.59 %	602000	Treacher-Collins syndrome 4, 618939 (3), Autosomal dominant
POLR1C	100 %	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR1D	99.99 %	613715	Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant
POLR3A	99.79 %	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	99.7 %	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant
POLR3GL	98.27 %	617457	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234 (3), Autosomal recessive
POP1	99.89 %	602486	Anauxetic dysplasia 2, 617396 (3), Autosomal recessive
POR	99.46 %	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
PORCN	99.81 %	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
PPIB	99.91 %	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive
PPP1CB	99.51 %	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PRKACA	99.74 %	601639	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (3); Cardioacrofacial dysplasia 1, 619142 (3), Autosomal dominant
PRKAR1A	96.1 %	188830	Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant

Skeletal dysplasia

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PRKG2	99.81 %	601591	Spondylometaphyseal dysplasia, Pagnamenta type, 619638 (3), Autosomal recessive; Acromesomelic dysplasia 4, 619636 (3), Autosomal recessive
PRMT7	99.65 %	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
PRRX1	99.21 %	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant
PSAT1	99.75 %	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSPH	98.61 %	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
PTCH1	99.87 %	601309	Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTDSS1	99.94 %	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
PTH1R	99.34 %	168468	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant; Eiken syndrome, 600002 (3), Autosomal recessive; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive
PTHLH	99.47 %	168470	Brachydactyly, type E2, 613382 (3), Autosomal dominant
PTPN11	99.84 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTPRD	99.88 %	601598	*No OMIM phenotypes*
PUF60	99.86 %	604819	Verheij syndrome, 615583 (3), Autosomal dominant
PYCR1	99.75 %	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
RAB23	99.9 %	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAB33B	99.98 %	605950	Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive
RAD21	99.87 %	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive
RASGRP2	99.76 %	605577	?Bleeding disorder, platelet-type, 18, 615888 (3), Autosomal recessive
RBBP8	99.93 %	604124	Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3)
RBM8A	99.16 %	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
RBPJ	99.83 %	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RECQL4	99.91 %	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
RFLNA	99.93 %	615927	*No OMIM phenotypes*
RFT1	98.55 %	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RIN1	99.61 %	605965	*No OMIM phenotypes*
RINT1	99.97 %	610089	Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive
RIPPLY2	99.74 %	609891	?Spondylocostal dysostosis 6, 616566 (3), Autosomal recessive
RMRP	98.59 %	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
RNU12	0 %	620204	CDAGS syndrome, 603116 (3), Autosomal recessive; ?Spinocerebellar ataxia, autosomal recessive 33, 620208 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RNU4ATAC	99.32 %	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
ROR2	99.79 %	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RPGRIP1L	96.51 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
RPL13	99.89 %	113703	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728 (3), Autosomal dominant
RSPO2	99.8 %	610575	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 (3), Autosomal recessive; Tetraamelia syndrome 2, 618021 (3), Autosomal recessive
RSPRY1	99.51 %	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
RUNX2	99.9 %	600211	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, 119600 (3), Autosomal dominant
SALL1	99.96 %	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
SALL4	99.99 %	607343	?IVIC syndrome, 147750 (3), Autosomal dominant; Duane-radial ray syndrome, 607323 (3), Autosomal dominant
SBDS	99.32 %	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SCARF2	98.63 %	613619	Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive
SCUBE3	99.44 %	614708	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 2, 619184 (3), Autosomal recessive
SDCCAG8	100 %	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SEC23A	99.88 %	610511	Craniolenticulosutural dysplasia, 607812 (3), Autosomal recessive, Autosomal dominant
SEC24D	99.62 %	607186	Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive
SERPINF1	99.91 %	172860	Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive
SERPINH1	99.67 %	600943	{Preterm premature rupture of the membranes, susceptibility to}, 610504 (3), Multifactorial; Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive
SETD2	99.71 %	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 70, 620157 (3), Autosomal dominant; Rabin-Pappas syndrome, 620155 (3), Autosomal dominant
SF3B4	99.47 %	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
SFRP4	99.94 %	606570	Pyle disease, 265900 (3), Autosomal recessive
SGK3	99.97 %	607591	*No OMIM phenotypes*
SGMS2	99.93 %	611574	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550 (3), Autosomal dominant
SGSH	99.79 %	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SH3BP2	99.54 %	602104	Cherubism, 118400 (3), Autosomal dominant
SH3PXD2B	99.96 %	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SHH	99.93 %	600725	Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia/coloboma 5, 611638 (3), Autosomal dominant
SHOX	95.24 %	312865	Short stature, idiopathic familial, 300582 (3); Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant; Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive
SIK3	99.88 %	614776	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162 (3), Autosomal recessive
SIX1	99.98 %	601205	Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant; Branchiootic syndrome 3, 608389 (3), Autosomal dominant
SIX2	99.96 %	604994	*No OMIM phenotypes*
SKI	98.92 %	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SLC10A7	99.96 %	611459	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive
SLC13A1	99.89 %	606193	*No OMIM phenotypes*
SLC17A5	99.7 %	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
SLC25A24	96.63 %	608744	Fontaine progeroid syndrome, 612289 (3), Autosomal dominant
SLC26A2	99.91 %	606718	Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive; De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive
SLC29A3	99.61 %	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC34A1	99.89 %	182309	?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant
SLC34A3	99.85 %	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
SLC35B2	99.96 %	610788	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269 (3), Autosomal recessive
SLC35C1	99.95 %	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC35D1	97.02 %	610804	Schneckenbecken dysplasia, 269250 (3), Autosomal recessive
SLC39A13	99.3 %	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive
SLC4A2	99.93 %	109280	?Osteopetrosis, autosomal recessive 9, 620366 (3), Autosomal recessive
SLCO2A1	99.75 %	601460	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 (3), Autosomal dominant; PHOAR2-enteropathy syndrome, 614441 (3), Autosomal recessive
SLCO5A1	99.84 %	613543	*No OMIM phenotypes*
SMAD2	99.89 %	601366	Loeys-Dietz syndrome 6, 619656 (3), Autosomal dominant; Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657 (3), Autosomal dominant
SMAD3	99.95 %	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
SMAD4	99.95 %	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SMAD6	99.91 %	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SMARCA4	99.81 %	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; ?Otosclerosis 12, 620792 (3), Autosomal dominant
SMARCAL1	99.78 %	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SMARCB1	99.8 %	601607	Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant
SMARCE1	99.26 %	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
SMC1A	99.93 %	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant
SMC3	99.92 %	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SMCHD1	99.84 %	614982	Facioscapulohumeral muscular dystrophy 2, digenic, 158901 (3), Digenic dominant; Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant
SMO	99.78 %	601500	Pallister-Hall-like syndrome, 241800 (3), Autosomal recessive; Basal cell carcinoma, somatic, 605462 (3); Curry-Jones syndrome, somatic mosaic, 601707 (3)
SMOC1	99.87 %	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMS	99.74 %	300105	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive
SNRPB	99 %	182282	Cerebrocostomandibular syndrome, 117650 (3), Autosomal dominant
SNX10	99.97 %	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
SOST	98.76 %	605740	Sclerosteosis 1, 269500 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant
SOX9	99.94 %	608160	Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant; 46XY sex reversal 10, 616425 (3), Autosomal dominant; Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant; 46XX sex reversal 2, 278850 (3), Autosomal dominant
SP7	100 %	606633	Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive
SPARC	98.85 %	182120	Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive
SPECC1L	99.96 %	614140	Teebi hypertelorism syndrome 1, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant
SQSTM1	99.97 %	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
SRP54	99.84 %	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
STAT3	99.7 %	102582	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
STT3A	99.92 %	601134	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 (3), Autosomal dominant; Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 (3), Autosomal recessive
SUCO	97.82 %	619434	*No OMIM phenotypes*
SULF1	99.89 %	610012	*No OMIM phenotypes*
SUMF1	99.88 %	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUZ12	98.21 %	606245	Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant
TAB2	99.63 %	605101	Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant
TALDO1	99.87 %	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
TAPT1	99.68 %	612758	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive
TBC1D24	99.87 %	613577	Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive
TBCE	99.8 %	604934	Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive
TBX15	98.52 %	604127	Cousin syndrome, 260660 (3), Autosomal recessive
TBX2	99.37 %	600747	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223 (3), Autosomal dominant
TBX3	99.88 %	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
TBX4	99.72 %	601719	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant; Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 (3), Autosomal recessive
TBX5	99.92 %	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
TBX6	99.57 %	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant
TBXAS1	99.89 %	274180	Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive
TCF12	99.93 %	600480	Craniosynostosis 3, 615314 (3), Autosomal dominant; Hypogonadotropic hypogonadism 26 with or without anosmia, 619718 (3), Autosomal recessive, Autosomal dominant
TCIRG1	99.78 %	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
TCOF1	99.76 %	606847	Treacher Collins syndrome 1, 154500 (3), Autosomal dominant
TCTN1	99.84 %	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	99.95 %	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	99.84 %	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TENT5A	99.95 %	611357	Osteogenesis imperfecta, type XVIII, 617952 (3), Autosomal recessive
TERT	99.86 %	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TFAP2B	99.8 %	601601	Patent ductus arteriosus 2, 617035 (3), Autosomal dominant; Char syndrome, 169100 (3), Autosomal dominant
TGDS	99.96 %	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
TGFB1	99.88 %	190180	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive; Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive
TGFB2	99.85 %	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant; Camurati-Engelmann disease 2, 606631 (3), Autosomal dominant
TGFB3	99.97 %	190230	Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant
TGFBR1	99.84 %	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	99.91 %	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
THPO	99.96 %	600044	Thrombocythemia 1, 187950 (3), Autosomal dominant; Thrombocytopenia 9, 620478 (3), Autosomal dominant; Amegakaryocytic thrombocytopenia, congenital, 2, 620481 (3), Autosomal recessive
TICRR	99.85 %	613298	*No OMIM phenotypes*
TLK2	97.61 %	608439	Intellectual developmental disorder, autosomal dominant 57, 618050 (3), Autosomal dominant
TMCO1	98.23 %	614123	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive
TMEM138	99.94 %	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM165	99.89 %	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
TMEM216	99.5 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Retinitis pigmentosa 98, 620996 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM231	93.22 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	99.12 %	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM251	99.97 %	619332	Dysostosis multiplex, Ain-Naz type, 619345 (3), Autosomal recessive
TMEM263	99.9 %		*No OMIM phenotypes*
TMEM38B	99.82 %	611236	Osteogenesis imperfecta, type XIV, 615066 (3), Autosomal recessive
TMEM53	99.66 %	619722	Craniotubular dysplasia, Ikegawa type, 619727 (3), Autosomal recessive
TMEM67	99.79 %	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TNFRSF11A	99.9 %	603499	Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteolysis, familial expansile, 174810 (3), Autosomal dominant
TNFRSF11B	99.75 %	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
TNFSF11	99.68 %	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
TOMM7	100 %	607980	Garg-Mishra progeroid syndrome, 620601 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TONSL	99.87 %	604546	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510 (3), Autosomal recessive
TOP2B	99.75 %	126431	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296 (3), Autosomal dominant
TP63	99.8 %	603273	Premature ovarian failure 21, 620311 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant
TRAF3IP1	99.94 %	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
TRAF7	99.27 %	606692	Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant
TRAIP	99.82 %	605958	Seckel syndrome 9, 616777 (3), Autosomal recessive
TRAPPC2	99.47 %	300202	Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive
TREM2	99.99 %	605086	{Alzheimer disease 17, susceptibility to}, 615080 (3), Autosomal recessive; Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal recessive
TRIM32	99.99 %	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRIP11	99.89 %	604505	Odontochondrodysplasia 1, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive
TRPS1	99.98 %	604386	Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant
TRPV4	99.9 %	605427	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); SED, Maroteaux type, 184095 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant
TRPV6	99.85 %	606680	Hyperparathyroidism, transient neonatal, 618188 (3), Autosomal recessive
TTC21B	99.13 %	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant
TTC8	99.76 %	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TWIST1	100 %	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant
TWIST2	99.99 %	607556	Ablepharon-macrostomia syndrome, 200110 (3), Autosomal dominant; Barber-Say syndrome, 209885 (3), Autosomal dominant; Focal facial dermal dysplasia 3, Setleis type, 227260 (3), Autosomal recessive
TXNL4A	99.98 %	611595	Burn-McKeown syndrome, 608572 (3), Autosomal recessive

Skeletal dysplasia

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TYROBP	99.88 %	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
UBA2	99.78 %	613295	ACCES syndrome, 619959 (3), Autosomal dominant
UFSP2	99.63 %	611482	?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant; Developmental and epileptic encephalopathy 106, 620028 (3), Autosomal recessive
UNC45A	99.78 %	611219	Osteootohepatoenteric syndrome, 619377 (3), Autosomal recessive
USP48	96.36 %	617445	Deafness, autosomal dominant 85, 620227 (3), Autosomal dominant
VAC14	99.53 %	604632	Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive
VCP	99.89 %	601023	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant
VDR	99.61 %	601769	Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
VPS33A	99.08 %	610034	Mucopolysaccharidosis-plus syndrome, 617303 (3), Autosomal recessive
VPS35L	98.94 %	618981	Ritscher-Schinzel syndrome 3, 619135 (3), Autosomal recessive
WBP11	99.71 %	618083	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227 (3), Autosomal dominant
WDPCP	99.79 %	613580	Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR19	99.62 %	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WDR35	99.87 %	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
WNT1	98.74 %	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3), Autosomal dominant; Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
WNT10B	99.68 %	601906	Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant; Split-hand/foot malformation 6, 225300 (3), Autosomal recessive
WNT3	99.83 %	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
WNT3A	99.82 %	606359	*No OMIM phenotypes*
WNT5A	99.71 %	164975	Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant
WNT6	99.71 %	604663	*No OMIM phenotypes*
WNT7A	99.91 %	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive; ?Santos syndrome, 613005 (3), Autosomal recessive
XRCC4	99.87 %	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
XYLT1	99.79 %	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
XYLT2	99.69 %	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
YY1	99.97 %	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant

Skeletal dysplasia

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ZEB2	99.86 %	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
ZIC1	99.95 %	600470	?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant
ZIC3	99.87 %	300265	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
ZMPSTE24	97.94 %	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive
ZNF462	99.95 %	617371	Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant
ZNF687	99.94 %	610568	Paget disease of bone 6, 616833 (3), Autosomal dominant
ZSWIM6	99.51 %	615951	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.