Skeletal_dysplasia panel

versie	v3 (450 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ABCC9	601439	Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant
ABL1	189980	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation; Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant
ACAN	155760	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; ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3), Autosomal dominant
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACVR1	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ADAMTS10	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
ADAMTS17	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
ADAMTSL2	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
AFF4	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
AGA	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
AHDC1	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
AKT1	164730	Breast cancer, somatic, 114480 (3); Cowden syndrome 6, 615109 (3); Proteus syndrome, somatic, 176920 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3)
ALG12	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
ALG3	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
ALG9	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive

ALPL	171760	Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant; Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive
ALX1	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
ALX3	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
ALX4	605420	Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant
AMER1	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
ANAPC1	608473	Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive
ANKH	605145	Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant; Chondrocalcinosis 2, 118600 (3), Autosomal dominant
ANKRD11	611192	KBG syndrome, 148050 (3), Autosomal dominant
ANO5	608662	Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant
ANTXR2	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
ARHGAP31	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARID1A	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
ARSL (ARSE)	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ASXL1	612990	Bohring-Opitz syndrome, 605039 (3), Autosomal dominant; Myelodysplastic syndrome, somatic, 614286 (3)
ASXL2	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
ATP6V0A2	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
B3GALT6	615291	Al-Gazali syndrome, 609465 (3); 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
B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
B3GLCT	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive

B4GALT7	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
B9D1	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
BBS4	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BHLHA9	615416	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3), Autosomal recessive; ?Camptosynpolydactyly, complex, 607539 (3), Autosomal recessive
BMP1	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
BMP2	112261	Brachydactyly, type A2, 112600 (3), Autosomal dominant; Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive
BMPER	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
BMPR1B	603248	Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant; Acromesomelic dysplasia, Demirhan type, 609441 (3), Autosomal recessive
BPNT2 (IMPAD1)	614010	Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive
C2CD3	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CANT1	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive
CASR	601199	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant; {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant

CC2D2A	612013	Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive
CCDC134	618788	No OMIM phenotype
CCDC8	614145	3-M syndrome 3, 614205 (3), Autosomal recessive
CCN6 (WISP3)	603400	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 (3), Autosomal recessive; Arthropathy, progressive pseudorheumatoid, of childhood, 208230 (3), Autosomal recessive
CCNQ	300708	STAR syndrome, 300707 (3), X-linked dominant
CD96	606037	C syndrome, 211750 (3), Autosomal dominant
CDC45	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
CDC6	602627	?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
CDH3	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
CDKN1C	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CDT1	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
CEP120	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive
CEP290	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive
CFAP410	603191	Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive; Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive
CHD7	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive
CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive
CHSY1	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
CILK1 (ICK)	612325	Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant
CKAP2L	616174	Filippi syndrome, 272440 (3), Autosomal recessive

CLCN5	300008	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Dent disease, 300009 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive
CLCN7	602727	Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant
COG1	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
COG4	606976	Saul-Wilson syndrome, 618150 (3), Autosomal dominant; Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive
COL10A1	120110	Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant
COL11A1	120280	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); Fibrochondrogenesis 1, 228520 (3), Autosomal recessive
COL11A2	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	120150	Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; {Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant
COL1A2	120160	{Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant

COL2A1	120140	Vitreoretinopathy with phalangeal epiphyseal dysplasia (3); Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Stickler sydrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant
COL9A1	120210	?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant; Stickler syndrome, type IV, 614134 (3)
COL9A2	120260	?Stickler syndrome, type V, 614284 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant
COL9A3	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; {Intervertebral disc disease, susceptibility to}, 603932 (3)
COLEC11	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
COMP	600310	Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant; Pseudoachondroplasia, 177170 (3), Autosomal dominant
CPLANE1	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
CREB3L1	616215	Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive
CREBBP	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant; Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant
CRTAP	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
CSF1R	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant
CSGALNACT1	616615	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870 (3), Autosomal recessive
CSPP1	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CTSA	613111	Galactosialidosis, 256540 (3), Autosomal recessive

CTSC	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive
CTSK	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
CUL7	609577	3-M syndrome 1, 273750 (3), Autosomal recessive
CYP27B1	609506	Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive
CYP2R1	608713	Rickets due to defect in vitamin D 25-hydroxylation, 600081 (3), Autosomal recessive
DCC	120470	Esophageal carcinoma, somatic, 133239 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive; Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3)
DDR2	191311	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive; Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant
DDX58	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
DDX59	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DHCR24	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHODH	126064	Miller syndrome, 263750 (3), Autosomal recessive
DIP2C	611380	No OMIM phenotype
DLL3	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive
DLL4	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DLX3	600525	Trichodontoosseous syndrome, 190320 (3), Autosomal dominant; Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant
DLX5	600028	Split-hand/foot malformation 1, 183600 (3), Autosomal dominant; ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
DLX6	600030	No OMIM phenotype
DMP1	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
DNMT3A	602769	Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant
DOCK6	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DPAGT1	191350	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
DPM1	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive

DSPP	125485	Dentin dysplasia, type II, 125420 (3), Autosomal dominant; Deafness, autosomal dominant 39, with dentinogenesis, 605594 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type II, 125490 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type III, 125500 (3), Autosomal dominant
DVL1	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
DVL3	601368	Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant
DYM	607461	Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
DYNC2I1 (WDR60)	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
DYNC2I2 (WDR34)	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
DYNC2LI1	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
DYNLT2B (TCTEX1D2)	617353	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive
EBP	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
EDNRA	131243	Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant; {Migraine, resistance to}, 157300 (3), Autosomal dominant
EFNA4	601380	No OMIM phenotype
EFNB1	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
EFTUD2	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EIF2AK3	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
EIF4A3	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
ENPP1	173335	Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
EOGT	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive

EP300	602700	Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant; Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3)
ERCC4	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive
ERF	611888	Craniosynostosis 4, 600775 (3), Autosomal dominant; Chitayat syndrome, 617180 (3), Autosomal dominant
ESCO2	609353	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (3), Autosomal recessive
EVC	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	607261	Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant; Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive
EXT1	608177	Exostoses, multiple, type 1, 133700 (3), Autosomal dominant; Chondrosarcoma, 215300 (3), Somatic mutation
EXT2	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
EXTL3	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
EZH2	601573	Weaver syndrome, 277590 (3), Autosomal dominant
FAM111A	615292	Gracile bone dysplasia, 602361 (3), Autosomal dominant; Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant
FAM20C	611061	Raine syndrome, 259775 (3), Autosomal recessive
FBLN1	135820	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4), Autosomal dominant
FBN1	134797	Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant
FBN2	612570	Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant; Macular degeneration, early-onset, 616118 (3), Autosomal dominant
FBXW4	608071/246560	-/Split-hand/foot malformation 3, gene duplication syndrome, 246560 (4), Autosomal dominant
FERMT3	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive

FGF10	602115	Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant
FGF16	300827	Metacarpal 4-5 fusion, 309630 (3), X-linked recessive
FGF23	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3), Autosomal recessive; Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant
FGF3	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
FGF4	164980	No OMIM phenotype
FGF8	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGF9	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant
FGFR1	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FGFR2	176943	Apert syndrome, 101200 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3); Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant
FGFR3	134934	Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Spermatocytic seminoma, somatic, 273300

		(3); Cervical cancer, somatic, 603956 (3); SADDAN, 616482 (3), Autosomal dominant
FIG4	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant
FKBP10	607063	Bruck syndrome 1, 259450 (3), Autosomal recessive; Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive
FLNA	300017	Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
FLNB	603381	Larsen syndrome, 150250 (3), Autosomal dominant; Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Boomerang dysplasia, 112310 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive; Atelosteogenesis, type III, 108721 (3), Autosomal dominant
FMN1	136535	No OMIM phenotype
FREM1	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant; Bifid nose with or without anorectal and renal anomalies, 608980 (3)
FUCA1	612280	Fucosidosis, 230000 (3), Autosomal recessive
FZD2	600667	Omodysplasia 2, 164745 (3), Autosomal dominant
GALNS	612222	Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive
GALNT3	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive

GDF3	606522	Microphthalmia, isolated 7, 613704 (3), Autosomal dominant; Microphthalmia with coloboma 6, 613703 (3), Autosomal dominant; Klippel-Feil syndrome 3, autosomal dominant, 613702 (3)
GDF5	601146	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3), Autosomal recessive; Symphalangism, proximal, 1B, 615298 (3), Autosomal dominant; Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant; Chondrodysplasia, Grebe type, 200700 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Du Pan syndrome, 228900 (3), Autosomal recessive; {Osteoarthritis-5}, 612400 (3); Brachydactyly, type C, 113100 (3), Autosomal dominant; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant
GDF6	601147	Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3)
GFER	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3)
GJA1	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant
GLB1	611458	GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLI3	165240	Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; {Hypothalamic hamartomas, somatic}, 241800 (3); Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant

GNAS	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPNAT1	616510	No OMIM phenotype
GNPTAB	607840	Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive
GNPTG	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GNS	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GORAB	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive
GPC6	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
GPX4	138322	Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive
GREM1	603054	No OMIM phenotype
GUSB	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
GZF1	613842	Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive
HDAC4	605314/600430	-/Chromosome 2q37 deletion syndrome, 600430 (4), Autosomal dominant
HDAC8	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HES7	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
HGSNAT	610453	Retinitis pigmentosa 73, 616544 (3), Autosomal recessive; Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive
HNRNPK	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
HOXA11	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant
HOXA13	142959	?Guttmacher syndrome, 176305 (3), Autosomal dominant; Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant
HOXD13	142989	Brachydactyly, type D, 113200 (3), Autosomal dominant; Brachydactyly, type E, 113300 (3), Autosomal dominant; ?Brachydactyly-syndactyly syndrome, 610713 (3); Syndactyly, type V, 186300 (3), Autosomal dominant; Synpolydactyly 1, 186000 (3), Autosomal dominant

HPGD	601688	Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; ?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive
HS2ST1	604844	No OMIM phenotype
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
IDH1	147700	{Glioma, susceptibility to, somatic}, 137800 (3)
IDH2	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDS	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IDUA	252800	Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive; Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive
IFIH1	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFITM5	614757	Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant
IFT122	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
IFT172	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT43	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive
IFT52	617094	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive
IFT80	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IFT81	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
IHH	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant
IKBKG	300248	Immunodeficiency 33, 300636 (3), X-linked recessive; Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive
IL11RA	600939	Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive

IL1RN	147679	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
INPPL1	600829	Opsismodysplasia, 258480 (3), Autosomal recessive
KAT6B	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant
KDELR2	No OMIM gene	No OMIM phenotype
KIAA0753	617112	?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive
KIAA1217	617367	No OMIM phenotype
KIF22	603213	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3), Autosomal dominant
KIF7	611254	?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
KMT2D	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
LBR	600024	Pelger-Huet anomaly, 169400 (3), Autosomal dominant; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Pelger-Huet anomaly with mild skeletal anomalies, 618019 (3)
LEMD3	607844	Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant; Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant
LFNG	602576	Spondylocostal dysostosis 3, autosomal recessive, 609813 (3), Autosomal recessive
LIFR	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
LMBR1	605522	Triphalangeal thumb-polysyndactyly syndrome, 174500 (3), Autosomal dominant; Syndactyly, type IV, 186200 (3), Autosomal dominant; Triphalangeal thumb, type I, 174500 (3), Autosomal dominant; Acheiropody, 200500 (3), Autosomal recessive; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Hypoplastic or aplastic tibia with polydactyly, 188740 (3), Autosomal dominant; Polydactyly, preaxial type II, 174500 (3), Autosomal dominant

LMNA	150330	Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Malouf syndrome, 212112 (3), Autosomal dominant
LMX1B	602575	Nail-patella syndrome, 161200 (3), Autosomal dominant
LONP1	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LPIN2	605519	Majeed syndrome, 609628 (3)
LRP4	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	603506	van Buchem disease, type 2, 607636 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant
LTBP2	602091	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; Glaucoma 3, primary congenital, D, 613086 (3); ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive
MAFB	608968	Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant
MAN2B1	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAN2C1	154580	No OMIM phenotype
MAP3K7	602614	Cardiospondylocarpofacial syndrome, 157800 (3), Autosomal dominant; Frontometaphyseal dysplasia 2, 617137 (3), Autosomal dominant
MASP1	600521	3MC syndrome 1, 257920 (3), Autosomal recessive

MATN3	602109	{Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, 608728 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant
MBTPS2	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
MEGF8	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEOX1	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
MESP2	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive
MGP	154870	Keutel syndrome, 245150 (3), Autosomal recessive
MIA2	602132	No OMIM phenotype
MIA3	613455	No OMIM phenotype
MKKS	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
MMP13	600108	Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive; Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant
MMP2	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
MMP9	120361	Metaphyseal anadysplasia 2, 613073 (3)
MNX1	142994	Currarino syndrome, 176450 (3), Autosomal dominant
MPDU1	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MSX2	123101	Parietal foramina 1, 168500 (3), Autosomal dominant; Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant
MTX2	608555	No OMIM phenotype
MYCN	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
MYLPF	617378	No OMIM phenotype
NAGLU	609701	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant
NANS	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive

NBAS	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
NEK1	604588	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant; Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive
NEU1	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
NF1	613113	Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant
NFIX	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant
NIPBL	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKX3-2	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive
NLRP3	606416	Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant; CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant
NOG	602991	Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant; Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant; Brachydactyly, type B2, 611377 (3), Autosomal dominant
NOTCH1	190198	Aortic valve disease 1, 109730 (3), Autosomal dominant; Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant
NOTCH2	600275	Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant; Alagille syndrome 2, 610205 (3), Autosomal dominant
NPPC	600296	No OMIM phenotype
NPR2	108961	Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant; Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Acromesomelic dysplasia, Maroteaux type, 602875 (3), Autosomal recessive
NSD1	606681	Sotos syndrome 1, 117550 (3), Autosomal dominant
NSDHL	300275	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive

NXN	612895	Robinow syndrome, autosomal recessive 2, 618529 (3), Autosomal recessive
OBSL1	610991	3-M syndrome 2, 612921 (3), Autosomal recessive
OFD1	300170	Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Joubert syndrome 10, 300804 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
ORC1	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
ORC4	603056	Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive
ORC6	607213	Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive
OSTM1	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
P3H1	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
P4HB	176790	Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant
PAM16	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive
PAN2	617447	No OMIM phenotype
PAPSS2	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
PAX3	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant
PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PCYT1A	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
PDE3A	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
PDE4D	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
PEX5	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHEX	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant

PHGDH	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
PIK3C2A	603601	Oculoskeletodental syndrome, 618440 (3), Autosomal recessive
PIK3CA	171834	Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); CLAPO syndrome, somatic, 613089 (3); Cowden syndrome 5, 615108 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Macrodactyly, somatic, 155500 (3); Keratosis, seborrheic, somatic, 182000 (3); Gastric cancer, somatic, 613659 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Nevus, epidermal, somatic, 162900 (3); CLOVE syndrome, somatic, 612918 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
PISD	612770	Liberfarb syndrome, 618889 (3), Autosomal recessive
PITX1	602149	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 (3), Autosomal dominant; Liebenberg syndrome, 186550 (4), Autosomal dominant
PLEKHM1	611466	Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant; ?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive
PLOD2	601865	Bruck syndrome 2, 609220 (3), Autosomal recessive
PLS3	300131	Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant
POC1A	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POLR1A	616404	Acrofacial dysostosis, Cincinnati type, 616462 (3), Autosomal dominant
POLR1C	610060	Treacher Collins syndrome 3, 248390 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive
POLR1D	613715	Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant
POP1	602486	Anauxetic dysplasia 2, 617396 (3), Autosomal recessive
POR	124015	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3); Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive
PPIB	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive

PRKAR1A	188830	Adrenocortical tumor, somatic (3); Myxoma, intracardiac, 255960 (3), Autosomal dominant; Carney complex, type 1, 160980 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant
PRKG2	601591	No OMIM phenotype
PRMT7	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
PSAT1	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSPH	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
PTDSS1	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
PTH1R	168468	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Eiken syndrome, 600002 (3), Autosomal recessive; Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive
PTHLH	168470	Brachydactyly, type E2, 613382 (3), Autosomal dominant
PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PUF60	604819	Verheij syndrome, 615583 (3), Autosomal dominant
PYCR1	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3); Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
RAB23	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAB33B	605950	Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive
RAD21	606462	?Mungan syndrome, 611376 (3), Autosomal recessive; Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant
RASGRP2	605577	?Bleeding disorder, platelet-type, 18, 615888 (3), Autosomal recessive
RBM8A	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
RBPJ	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RECQL4	603780	RAPADILINO syndrome, 266280 (3), Autosomal recessive; Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2,, 268400 (3), Autosomal recessive
RFT1	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RIN1	No OMIM gene	No OMIM phenotype

RNU4ATAC	601428	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive; Roifman syndrome, 616651 (3), Autosomal recessive
ROR2	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RPGRIP1L	610937	COACH syndrome, 216360 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive
RPL13	113703	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728 (3), Autosomal dominant
RUNX2	600211	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant; Cleidocranial dysplasia, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant
SALL1	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
SALL4	607343	Duane-radial ray syndrome, 607323 (3), Autosomal dominant; IVIC syndrome, 147750 (3), Autosomal dominant
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SCARF2	613619	Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive
SCUBE3	614708	No OMIM phenotype
SEC24D	607186	Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive
SERPINF1	172860	Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive
SERPINH1	600943	Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive; {Preterm premature rupture of the membranes, susceptibility to}, 610504 (3)
SETD2	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
SF3B4	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
SFRP4	606570	Pyle disease, 265900 (3), Autosomal recessive
SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SH3BP2	602104	Cherubism, 118400 (3), Autosomal dominant
SH3PXD2B	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive
SHH	600725	Schizencephaly, 269160 (3); Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant

SHOX	400020	Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant; Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive; Short stature, idiopathic familial, 300582 (3)
SKI	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SLC10A7	611459	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive
SLC17A5	604322	Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive; Salla disease, 604369 (3), Autosomal recessive
SLC26A2	606718	De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Achondrogenesis Ib, 600972 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive
SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC34A1	182309	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; ?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive
SLC34A3	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
SLC35C1	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC35D1	610804	Schneckenbecken dysplasia, 269250 (3), Autosomal recessive
SLC39A13	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive
SLCO2A1	601460	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441 (3), Autosomal recessive
SLCO5A1	613543	No OMIM phenotype
SMAD3	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
SMAD4	600993	Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3)
SMARCAL1	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SMC1A	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 85, with or without midline brain defects, 301044 (3)
SMC3	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant

SMO	601500	Curry-Jones syndrome, somatic mosaic, 601707 (3); Pallister-Hall-like syndrome, 241800 (3), Autosomal recessive; Basal cell carcinoma, somatic, 605462 (3)
SMOC1	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SNRPB	182282	Cerebrocostomandibular syndrome, 117650 (3), Autosomal dominant
SNX10	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
SOST	605740	Sclerosteosis 1, 269500 (3), Autosomal recessive; Van Buchem disease, 239100 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant
SOX9	608160	Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant
SP7	606633	Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive
SPARC	182120	Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive
SUCO	No OMIM gene	No OMIM phenotype
SULF1	610012	No OMIM phenotype
SUMF1	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
TALDO1	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
TAPT1	612758	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive
TBCE	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	604127	Cousin syndrome, 260660 (3), Autosomal recessive
TBX3	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
TBX4	601719	Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 (3), Autosomal recessive; Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant
TBX5	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
TBX6	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant
TBXAS1	274180	Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive
TCF12	600480	Craniosynostosis 3, 615314 (3), Autosomal dominant
TCIRG1	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive

TCOF1	606847	Treacher Collins syndrome 1, 154500 (3), Autosomal dominant
TCTN2	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	613847	Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive; Joubert syndrome 18, 614815 (3), Autosomal recessive
TENT5A	611357	Osteogenesis imperfecta, type XVIII, 617952 (3), Autosomal recessive
TERT	187270	{Melanoma, cutaneous malignant, 9}, 615134 (3); {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation; {Dyskeratosis congenita, autosomal recessive 4}, 613989 (3), Autosomal recessive, Autosomal dominant; {Dyskeratosis congenita, autosomal dominant 2}, 613989 (3), Autosomal recessive, Autosomal dominant
TGFB1	190180	{Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive; Camurati-Engelmann disease, 131300 (3), Autosomal dominant; Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive
TGFB2	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant
TGFBR1	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
TGFBR2	190182	Esophageal cancer, somatic, 133239 (3); Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
THPO	600044	Thrombocythemia 1, 187950 (3), Autosomal dominant
TMEM165	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
TMEM216	613277	Meckel syndrome 2, 603194 (3), Autosomal recessive; Joubert syndrome 2, 608091 (3), Autosomal recessive
TMEM231	614949	Meckel syndrome 11, 615397 (3), Autosomal recessive; Joubert syndrome 20, 614970 (3), Autosomal recessive
TMEM38B	611236	Osteogenesis imperfecta, type XIV, 615066 (3)
TMEM67	609884	Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive
TNFRSF11A	603499	Osteolysis, familial expansile, 174810 (3), Autosomal dominant; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive

TNFRSF11B	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
TNFSF11	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
TONSL	604546	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510 (3), Autosomal recessive
TP63	603273	Limb-mammary syndrome, 603543 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant
TRAPPC2	300202	Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive
TREM2	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal dominant
TRIP11	604505	Osteochondrodysplasia, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive
TRPS1	604386	Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant
TRPV4	605427	Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; SED, Maroteaux type, 184095 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIII, 600175 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant
TRPV6	606680	Hyperparathyroidism, transient neonatal, 618188 (3), Autosomal recessive
TTC21B	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
TTC8	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TWIST1	601622	Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Craniosynostosis 1, 123100 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant

TYROBP	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
VDR	601769	Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
VPS35L	618981	No OMIM phenotype
WDPCP	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR19	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
WDR35	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
WNT1	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3); Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
WNT10B	601906	Split-hand/foot malformation 6, 225300 (3), Autosomal recessive; Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant
WNT3	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
WNT3A	606359	No OMIM phenotype
WNT5A	164975	Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant
WNT7A	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive
XRCC4	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
XYLT1	608124	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Desbuquois dysplasia 2, 615777 (3), Autosomal recessive
XYLT2	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
ZMPSTE24	606480	Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive
ZSWIM6	615951	Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant; Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Sep 01, 2020

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.