

Skeletal_dysplasia panel		
versie	V1 (345 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABCC9</i>	601439	Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 10, 608569 (3); Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant
<i>ABL1</i>	189980	Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant; Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation
<i>ACAN</i>	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
<i>ACP5</i>	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
<i>ACVR1</i>	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
<i>ADAMTS10</i>	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
<i>ADAMTS17</i>	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
<i>ADAMTSL2</i>	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
<i>AFF4</i>	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
<i>AGA</i>	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
<i>AGPS</i>	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
<i>AHDC1</i>	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
<i>AKT1</i>	164730	Breast cancer, somatic, 114480 (3); Colorectal cancer, somatic, 114500 (3); Cowden syndrome 6, 615109 (3); Ovarian cancer, somatic, 167000 (3); Proteus syndrome, somatic, 176920 (3); {Schizophrenia, susceptibility to}, 181500 (2), Autosomal dominant
<i>ALPL</i>	171760	Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant
<i>ALX1</i>	601527	?Frontonasal dysplasia 3, 613456 (3)
<i>ALX3</i>	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive

<i>ALX4</i>	605420	{Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant
<i>AMER1</i>	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
<i>ANKH</i>	605145	Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant
<i>ANOS</i>	608662	Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive
<i>ANTXR2</i>	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
<i>ARHGAP31</i>	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
<i>ARID1A</i>	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
<i>ARID1B</i>	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
<i>ARSB</i>	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
<i>ARSL (ARSE)</i>	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
<i>ATP6VOA2</i>	611716	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal recessive
<i>B3GALT6</i>	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
<i>B3GAT3</i>	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
<i>B4GALT7</i>	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
<i>BMP1</i>	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
<i>BMP2</i>	112261	Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive; Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 (3), Autosomal dominant
<i>BMPER</i>	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
<i>BMPR1B</i>	603248	Acromesomelic dysplasia, Demirhan type, 609441 (3), Autosomal recessive; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant
<i>CA2</i>	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
<i>CANT1</i>	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive

<i>CASR</i>	601199	{Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant
<i>CC2D2A</i>	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
<i>CCDC8</i>	614145	3-M syndrome 3, 614205 (3), Autosomal recessive
<i>CCN6</i> (<i>WISP3</i>)	603400	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 (3), Autosomal recessive; Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 (3), Autosomal recessive
<i>CCNQ</i> (<i>FAM58A</i>)	300708	STAR syndrome, 300707 (3), X-linked dominant
<i>CD96</i>	606037	C syndrome, 211750 (3), Autosomal dominant
<i>CDC45</i>	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
<i>CDC6</i>	602627	?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
<i>CDH3</i>	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
<i>CDKN1C</i>	600856	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
<i>CDT1</i>	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
<i>CEP120</i>	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive
<i>CEP290</i>	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive
<i>CHD7</i>	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
<i>CHST14</i>	608429	Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive
<i>CHST3</i>	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive
<i>CHSY1</i>	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
<i>CILK1</i> (<i>ICK</i>)	612325	Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant
<i>CKAP2L</i>	616174	Filippi syndrome, 272440 (3), Autosomal recessive

<i>CLCN5</i>	300008	Dent disease, 300009 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive; Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive
<i>CLCN7</i>	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive
<i>COG1</i>	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
<i>COL10A1</i>	120110	Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant
<i>COL11A1</i>	120280	?Deafness, autosomal dominant 37, 618533 (3); Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; {Lumbar disc herniation, susceptibility to}, 603932 (3); Marshall syndrome, 154780 (3), Autosomal dominant; Stickler syndrome, type II, 604841 (3), Autosomal dominant
<i>COL11A2</i>	120290	Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal recessive, Autosomal dominant; Otopondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant; Otopondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive {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 I, 166200 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant
<i>COL1A1</i>	120150	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant

		Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)
<i>COL2A1</i>	120140	
<i>COL9A1</i>	120210	?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant; Stickler syndrome, type IV, 614134 (3)
<i>COL9A2</i>	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
<i>COL9A3</i>	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; {Intervertebral disc disease, susceptibility to}, 603932 (3)
<i>COMP</i>	600310	Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant; Pseudoachondroplasia, 177170 (3), Autosomal dominant
<i>CPLANE1 (C5orf42)</i>	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
<i>CREB3L1</i>	616215	Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive
<i>CREBBP</i>	600140	Menke-Hennekam syndrome 1, 618332 (3); Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
<i>CRTAP</i>	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
<i>CTSA</i>	613111	Galactosialidosis, 256540 (3), Autosomal recessive
<i>CTSK</i>	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
<i>CUL7</i>	609577	3-M syndrome 1, 273750 (3), Autosomal recessive
<i>DDR2</i>	191311	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive; Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant
<i>DDX58</i>	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
<i>DDX59</i>	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
<i>DHCR24</i>	606418	Desmosterolosis, 602398 (3), Autosomal recessive
<i>DHODH</i>	126064	Miller syndrome, 263750 (3), Autosomal recessive
<i>DIP2C</i>	611380	No OMIM phenotype
<i>DLL3</i>	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive

<i>DLX3</i>	600525	Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant; Trichodontoosseous syndrome, 190320 (3), Autosomal dominant
<i>DLX5</i>	600028	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
<i>DLX6</i>	600030	No OMIM phenotype
<i>DMP1</i>	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
<i>DOCK6</i>	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
<i>DVL1</i>	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
<i>DYM</i>	607461	Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive; Smith-McCort dysplasia, 607326 (3), Autosomal recessive
<i>DYNC2H1</i>	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
<i>DYNC2LI1</i>	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
<i>EBP</i>	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
<i>EDNRA</i>	131243	Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant; {Migraine, resistance to}, 157300 (3), Autosomal dominant
<i>EFNA4</i>	601380	No OMIM phenotype
<i>EFNB1</i>	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
<i>EFTUD2</i>	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
<i>EIF2AK3</i>	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
<i>EIF4A3</i>	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
<i>ENPP1</i>	173335	Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>EOGT</i>	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
<i>EP300</i>	602700	Colorectal cancer, somatic, 114500 (3); Menke-Hennekam syndrome 2, 618333 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
<i>ERCC4</i>	133520	Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive

H9.1-OP2-B40: Genpanel Skeletal dysplasia, V1, in voege op 14/02/2020

<i>ERF</i>	611888	Chitayat syndrome, 617180 (3), Autosomal dominant; Craniosynostosis 4, 600775 (3), Autosomal dominant
<i>ESCO2</i>	609353	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (3), Autosomal recessive
<i>EVC</i>	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EVC2</i>	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EXT1</i>	608177	Chondrosarcoma, 215300 (3), Autosomal recessive; Exostoses, multiple, type 1, 133700 (3), Autosomal dominant
<i>EXT2</i>	608210	Exostoses, multiple, type 2, 133701 (3), Autosomal dominant; Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive
<i>EZH2</i>	601573	Weaver syndrome, 277590 (3), Autosomal dominant
<i>FAM111A</i>	615292	Gracile bone dysplasia, 602361 (3), Autosomal dominant; Kenny- Caffey syndrome, type 2, 127000 (3), Autosomal dominant
<i>FAM20C</i>	611061	Raine syndrome, 259775 (3), Autosomal recessive
<i>FBLN1</i>	135820	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4), Autosomal dominant
<i>FBN1</i>	134797	Acromicric dysplasia, 102370 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; MASS syndrome, 604308 (3); Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant
<i>FBN2</i>	612570	Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant; Macular degeneration, early-onset, 616118 (3), Autosomal dominant
<i>FBXW4</i>	608071	No OMIM phenotype
<i>FERMT3</i>	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
<i>FGF10</i>	602115	Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant
<i>FGF16</i>	300827	Metacarpal 4-5 fusion, 309630 (3), X-linked recessive
<i>FGF23</i>	605380	Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant; Osteomalacia, tumor-induced (1); Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3)
<i>FGF3</i>	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
<i>FGF4</i>	164980	No OMIM phenotype
<i>FGF8</i>	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
<i>FGF9</i>	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant

<i>FGFR1</i>	136350	Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3); Hartsfield syndrome, 615465 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Trignocephaly 1, 190440 (3), Autosomal dominant
<i>FGFR2</i>	176943	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Crouzon syndrome, 123500 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)
<i>FGFR3</i>	134934	Achondroplasia, 100800 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); SADDAN, 616482 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant
<i>FIG4</i>	609390	Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Yunis-Varon syndrome, 216340 (3), Autosomal recessive
<i>FKBP10</i>	607063	Bruck syndrome 1, 259450 (3), Autosomal recessive; Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive

<i>FLNA</i>	300017	Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; Congenital short bowel syndrome, 300048 (3), X-linked recessive; ?FG syndrome 2, 300321 (3), X-linked; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant
<i>FLNB</i>	603381	Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Boomerang dysplasia, 112310 (3), Autosomal dominant; Larsen syndrome, 150250 (3), Autosomal dominant; Spondylorcarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive
<i>FMN1</i>	136535	No OMIM phenotype
<i>FREM1</i>	608944	Bifid nose with or without anorectal and renal anomalies, 608980 (3); Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant
<i>FUCA1</i>	612280	Fucosidosis, 230000 (3), Autosomal recessive
<i>FZD2</i>	600667	Omodysplasia 2, 164745 (3), Autosomal dominant
<i>GALNS</i>	612222	Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive
<i>GALNT3</i>	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
<i>GDF3</i>	606522	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant
<i>GDF5</i>	601146	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type C, 113100 (3), Autosomal dominant; Chondrodysplasia, Grebe type, 200700 (3), Autosomal recessive; Du Pan syndrome, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Symphalangism, proximal, 1B, 615298 (3)
<i>GDF6</i>	601147	Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant
<i>GFER</i>	600924	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076 (3)

<i>GJA1</i>	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
<i>GLB1</i>	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive
<i>GLI3</i>	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; {Hypothalamic hamartomas, somatic}, 241800 (3); Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
<i>GNAS</i>	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; 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; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
<i>GNPAT</i>	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
<i>GNPTAB</i>	607840	Mucopolysaccharidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucopolysaccharidosis III alpha/beta, 252600 (3), Autosomal recessive
<i>GNPTG</i>	607838	Mucopolysaccharidosis III gamma, 252605 (3), Autosomal recessive
<i>GNS</i>	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
<i>GORAB</i>	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive
<i>GPC6</i>	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
<i>GPX4</i>	138322	Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive
<i>GREM1</i>	603054	No OMIM phenotype
<i>GUSB</i>	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
<i>HDAC4</i>	605314	No OMIM phenotype
<i>HDAC8</i>	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
<i>HES7</i>	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive

<i>HGSNAT</i>	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
<i>HOXA11</i>	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant
<i>HOXA13</i>	142959	?Guttmacher syndrome, 176305 (3), Autosomal dominant; Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant
<i>HOXD13</i>	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
<i>HPGD</i>	601688	Cranioosteoarthropathy, 259100 (3), Autosomal recessive; Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive
<i>HSPG2</i>	142461	Dysegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
<i>IDH1</i>	147700	{Glioma, susceptibility to, somatic}, 137800 (3)
<i>IDH2</i>	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
<i>IDS</i>	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
<i>IDUA</i>	252800	Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive
<i>IFIH1</i>	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IFITM5</i>	614757	Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant
<i>IFT122</i>	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
<i>IFT140</i>	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
<i>IFT172</i>	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
<i>IFT43</i>	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
<i>IFT80</i>	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
<i>IHH</i>	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant

<i>IKBKG</i>	300248	Ectodermal dysplasia and immunodeficiency 1, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)
<i>IL1RN</i>	147679	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive; {Microvascular complications of diabetes 4}, 612628 (3)
<i>IMPAD1</i>	614010	Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive
<i>INPPL1</i>	600829	Opsismodysplasia, 258480 (3), Autosomal recessive
<i>KAT6B</i>	605880	Genitopatellar syndrome, 606170 (3), Autosomal dominant; SBBYSS syndrome, 603736 (3), Autosomal dominant
<i>KIF22</i>	603213	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3), Autosomal dominant
<i>LBR</i>	600024	Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive; Pelger-Huet anomaly, 169400 (3), Autosomal dominant; Pelger-Huet anomaly with mild skeletal anomalies, 618019 (3); ?Reynolds syndrome, 613471 (3), Autosomal dominant
<i>LEMD3</i>	607844	Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant; Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant
<i>LFNG</i>	602576	Spondylocostal dysostosis 3, autosomal recessive, 609813 (3), Autosomal recessive
<i>LIFR</i>	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
<i>LMBR1</i>	605522	Acheiropody, 200500 (3), Autosomal recessive; Hypoplastic or aplastic tibia with polydactyly, 188740 (3), Autosomal dominant; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Polydactyly, preaxial type II, 174500 (3), Autosomal dominant; Syndactyly, type IV, 186200 (3), Autosomal dominant; Triphalangeal thumb, type I, 174500 (3), Autosomal dominant; Triphalangeal thumb-polysyndactyly syndrome, 174500 (3), Autosomal dominant

<i>LMNA</i>	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (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; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
<i>LMX1B</i>	602575	Nail-patella syndrome, 161200 (3), Autosomal dominant
<i>LONP1</i>	605490	CODAS syndrome, 600373 (3), Autosomal recessive
<i>LPIN2</i>	605519	Majeed syndrome, 609628 (3)
<i>LRP4</i>	604270	Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant
<i>LRP5</i>	603506	[Bone mineral density variability 1], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; {Osteoporosis}, 166710 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; van Buchem disease, type 2, 607636 (3), Autosomal dominant
<i>LTBP2</i>	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
<i>MAFB</i>	608968	Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MAN2C1</i>	154580	No OMIM phenotype
<i>MATN3</i>	602109	Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant; {Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, 608728 (3), Autosomal recessive

<i>MBTPS2</i>	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive
<i>MEGF8</i>	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
<i>MEOX1</i>	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
<i>MESP2</i>	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive
<i>MGP</i>	154870	Keutel syndrome, 245150 (3), Autosomal recessive
<i>MIA2 (CTAGE5)</i>	602132	No OMIM phenotype
<i>MIA3</i>	613455	No OMIM phenotype
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
<i>MMP13</i>	600108	Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant
<i>MMP2</i>	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
<i>MMP9</i>	120361	Metaphyseal anadysplasia 2, 613073 (3)
<i>MXN1</i>	142994	Currarino syndrome, 176450 (3), Autosomal dominant
<i>MSX2</i>	123101	Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina 1, 168500 (3), Autosomal dominant; Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant
<i>MYCN</i>	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
<i>NAGLU</i>	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
<i>NBAS</i>	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
<i>NEK1</i>	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
<i>NEU1</i>	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
<i>NF1</i>	613113	Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant

<i>NFIX</i>	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant
<i>NIPBL</i>	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
<i>NKX3-2</i>	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive
<i>NLRP3</i>	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
<i>NOG</i>	602991	Brachydactyly, type B2, 611377 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant
<i>NOTCH2</i>	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
<i>NPPC</i>	600296	No OMIM phenotype
<i>NPR2</i>	108961	Acromesomelic dysplasia, Maroteaux type, 602875 (3), Autosomal recessive; Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant
<i>NSD1</i>	606681	Leukemia, acute myeloid, 601626 (1), Autosomal dominant, Somatic mutation; Sotos syndrome 1, 117550 (3), Autosomal dominant
<i>NSDHL</i>	300275	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
<i>OBSL1</i>	610991	3-M syndrome 2, 612921 (3), Autosomal recessive
<i>OFD1</i>	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
<i>ORC1</i>	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
<i>ORC4</i>	603056	Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive
<i>ORC6</i>	607213	Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive
<i>OSTM1</i>	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
<i>P3H1</i>	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
<i>P4HB</i>	176790	Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant
<i>PAM16</i>	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive
<i>PAN2</i>	617447	No OMIM phenotype

<i>PAPSS2</i>	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
<i>PCNT</i>	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
<i>PCYT1A</i>	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
<i>PDE3A</i>	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
<i>PDE4D</i>	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PHEX</i>	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
<i>PIGV</i>	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
<i>PIK3CA</i>	171834	Breast cancer, somatic, 114480 (3); CLAPO syndrome, somatic, 613089 (3); CLOVE syndrome, somatic, 612918 (3); Colorectal cancer, somatic, 114500 (3); Cowden syndrome 5, 615108 (3); Gastric cancer, somatic, 613659 (3); Hepatocellular carcinoma, somatic, 114550 (3); Keratosis, seborrheic, somatic, 182000 (3); Macrodactyly, somatic, 155500 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Nevus, epidermal, somatic, 162900 (3); Non-small cell lung cancer, somatic, 211980 (3); Ovarian cancer, somatic, 167000 (3)
<i>PITX1</i>	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
<i>PLEKHM1</i>	611466	Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant; ?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive
<i>PLOD2</i>	601865	Bruck syndrome 2, 609220 (3), Autosomal recessive
<i>PLS3</i>	300131	Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant
<i>POLR1C</i>	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
<i>POLR1D</i>	613715	Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant
<i>POP1</i>	602486	Anauxetic dysplasia 2, 617396 (3), Autosomal recessive
<i>POR</i>	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
<i>PPIB</i>	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive

<i>PRKAR1A</i>	188830	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; Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant
<i>PTDSS1</i>	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
<i>PTH1R</i>	168468	Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive; Eiken syndrome, 600002 (3), Autosomal recessive; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant
<i>PTH1H</i>	168470	Brachydactyly, type E2, 613382 (3), Autosomal dominant
<i>PTPN11</i>	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant
<i>PYCR1</i>	179035	Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIIB, 614438 (3)
<i>RAB23</i>	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
<i>RAB33B</i>	605950	Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive
<i>RAD21</i>	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive
<i>RASGRP2</i>	605577	?Bleeding disorder, platelet-type, 18, 615888 (3), Autosomal recessive
<i>RBM8A</i>	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
<i>RBPJ</i>	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
<i>RECQL4</i>	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive; Rothmund-Thomson syndrome, 268400 (3), Autosomal recessive
<i>RIN1</i>	No OMIM gene	No OMIM phenotype
<i>ROR2</i>	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
<i>RUNX2</i>	600211	Cleidocranial dysplasia, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant

<i>SALL1</i>	602218	Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant; Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant
<i>SALL4</i>	607343	Duane-radial ray syndrome, 607323 (3), Autosomal dominant; IVIC syndrome, 147750 (3), Autosomal dominant
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
<i>SEC24D</i>	607186	Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive
<i>SERPINF1</i>	172860	Osteogenesis imperfecta, type VI, 613982 (3)
<i>SERPINH1</i>	600943	Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive; {Preterm premature rupture of the membranes, susceptibility to}, 610504 (3)
<i>SETD2</i>	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
<i>SF3B4</i>	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
<i>SH3BP2</i>	602104	Cherubism, 118400 (3), Autosomal dominant
<i>SH3PXD2B</i>	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive
<i>SHH</i>	600725	Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant
<i>SHOX</i>	400020	Langer mesomelic dysplasia, 249700 (3); Leri-Weill dyschondrosteosis, 127300 (3); Short stature, idiopathic familial, 300582 (3)
<i>SKI</i>	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
<i>SLC17A5</i>	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<i>SLC26A2</i>	606718	Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (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; Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive
<i>SLC29A3</i>	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
<i>SLC34A3</i>	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
<i>SLC35D1</i>	610804	Schneckenbecken dysplasia, 269250 (3), Autosomal recessive
<i>SLC39A13</i>	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive
<i>SLCO5A1</i>	613543	No OMIM phenotype
<i>SMAD3</i>	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant

<i>SMAD4</i>	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
<i>SMARCAL1</i>	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
<i>SMC1A</i>	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant
<i>SMC3</i>	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
<i>SNX10</i>	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
<i>SOST</i>	605740	Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant; Sclerosteosis 1, 269500 (3), Autosomal recessive; Van Buchem disease, 239100 (3), Autosomal recessive
<i>SOX9</i>	608160	Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant
<i>SP7</i>	606633	Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive
<i>SUCO</i>	No OMIM gene	No OMIM phenotype
<i>SULF1</i>	610012	No OMIM phenotype
<i>SUMF1</i>	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<i>TBCE</i>	604934	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive
<i>TBX15</i>	604127	Cousin syndrome, 260660 (3), Autosomal recessive
<i>TBX3</i>	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
<i>TBX4</i>	601719	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant
<i>TBX5</i>	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
<i>TBX6</i>	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant
<i>TBXAS1</i>	274180	Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive; ?Thromboxane synthase deficiency, 614158 (1), Autosomal dominant
<i>TCF12</i>	600480	Craniosynostosis 3, 615314 (3), Autosomal dominant
<i>TCIRG1</i>	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
<i>TCOF1</i>	606847	Treacher Collins syndrome 1, 154500 (3), Autosomal dominant
<i>TCTN3</i>	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
<i>TGFB1</i>	190180	Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive; Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive

H9.1-OP2-B40: Genpanel Skeletal dysplasia, V1, in voege op 14/02/2020

<i>TGFB2</i>	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant
<i>TGFBR1</i>	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
<i>TGFBR2</i>	190182	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
<i>THPO</i>	600044	Thrombocythemia 1, 187950 (3), Autosomal dominant
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<i>TMEM38B</i>	611236	Osteogenesis imperfecta, type XIV, 615066 (3)
<i>TMEM67</i>	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; ?RHYS syndrome, 602152 (3), Autosomal recessive
<i>TNFRSF11A</i>	603499	Osteolysis, familial expansile, 174810 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant
<i>TNFRSF11B</i>	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
<i>TNFSF11</i>	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
<i>TP63</i>	603273	ADULT syndrome, 103285 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant
<i>TRAPPC2</i>	300202	Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive
<i>TREM2</i>	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3)
<i>TRIP11</i>	604505	Achondrogenesis, type IA, 200600 (3), Autosomal recessive; Osteochondrodysplasia, 184260 (3), Autosomal recessive
<i>TRPS1</i>	604386	Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant

<i>TRPV4</i>	605427	?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; SED, Maroteaux type, 184095 (3), Autosomal dominant; Scapulooperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); Spinal muscular atrophy, distal, congenital nonprogressive, 600175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant
<i>TTC21B</i>	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
<i>TWIST1</i>	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant
<i>TYROBP</i>	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
<i>WDR19</i>	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
<i>WDR34</i>	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
<i>WDR35</i>	613602	Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive; Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive
<i>WNT1</i>	164820	Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive; {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3)
<i>WNT10B</i>	601906	Split-hand/foot malformation 6, 225300 (3), Autosomal recessive; Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant
<i>WNT3</i>	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
<i>WNT3A</i>	606359	No OMIM phenotype
<i>WNT5A</i>	164975	Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant
<i>WNT7A</i>	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive

<i>XYLT1</i>	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
<i>XYLT2</i>	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
<i>ZMPSTE24</i>	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive

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: Sept 30, 2019

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.