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SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.2.0 (647 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
ABCC9	n.t.b					Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ACAN	n.t.b					?Spondyloepiphyseal dysplasia, Kimberley type, 608361;Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800;Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACP5	n.t.b					Spondyloenchondrodysplasia with immune dysregulation, 607944

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ACTB	n.t.b					Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ACTG1	n.t.b					Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ACVR1	n.t.b					Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	n.t.b					Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS15	n.t.b					Arthrogryposis, distal, type 12, 620545
ADAMTS17	n.t.b					Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	n.t.b					Geleophysic dysplasia 1, 231050
AFF3	n.t.b					KINSSHIP syndrome, 619297
AGA	n.t.b					Aspartylglucosaminuria, 208400
AGPS	n.t.b					Rhizomelic chondrodysplasia punctata, type 3, 600121

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AIFM1	n.t.b					Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
ALG12	n.t.b					Congenital disorder of glycosylation, type Ig, 607143
ALG3	n.t.b					Congenital disorder of glycosylation, type Id, 601110
ALG9	n.t.b					Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALMS1	n.t.b					Alstrom syndrome, 203800
ALPL	n.t.b					Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300
ALX1	n.t.b					Frontonasal dysplasia 3, 613456
ALX3	n.t.b					Frontonasal dysplasia 1, 136760
ALX4	n.t.b					Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMER1	n.t.b					Osteopathia striata with cranial sclerosis, 300373

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AMMECR1	n.t.b					Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANAPC1	n.t.b					Rothmund-Thomson syndrome, type 1, 618625
ANKH	n.t.b					Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000
ANKRD11	n.t.b					KBG syndrome, 148050
ANO5	n.t.b					Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260
ANTXR2	n.t.b					Hyaline fibromatosis syndrome, 228600
APC2	n.t.b					Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
ARCN1	n.t.b					Short stature-micrognathia syndrome, 617164
ARHGAP31	n.t.b					Adams-Oliver syndrome 1, 100300
ARID1A	n.t.b					Coffin-Siris syndrome 2, 614607
ARID1B	n.t.b					Coffin-Siris syndrome 1, 135900
ARSB	n.t.b					Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200

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ARSK	n.t.b					Mucopolysaccharidosis, type X, 619698
ARSL	n.t.b					Chondrodysplasia punctata, X-linked recessive, 302950
ASXL1	n.t.b					Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ATP6V0A2	n.t.b					Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200
ATP7A	n.t.b					Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATR	n.t.b					Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN1	n.t.b					Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864
B3GALT6	n.t.b					Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465

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B3GAT3	n.t.b					Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	n.t.b					Peters-plus syndrome, 261540
B4GALT7	n.t.b					Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BANF1	n.t.b					Nestor-Guillermo progeria syndrome, 614008
BGN	n.t.b					Meester-Loeys syndrome, 300989;Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	n.t.b					?Camptosynpolydactyly, complex, 607539;Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BLM	n.t.b					Bloom syndrome, 210900
BMP1	n.t.b					Osteogenesis imperfecta, type XIII, 614856
BMP2	n.t.b					Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200
BMP5	n.t.b					
BMPER	n.t.b					Diaphanospondylodysostosis, 608022

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BMPR1B	n.t.b					Acromesomelic dysplasia 3, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849
BPNT2	n.t.b					Chondrodysplasia with joint dislocations, GPAPP type, 614078
BRAF	n.t.b					Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
BRF1	n.t.b					Cerebellofaciodental syndrome, 616202
BTK	n.t.b					Agammaglobulinemia, X-linked 1, 300755;Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	n.t.b					
BUB1B	n.t.b					Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
CA2	n.t.b					Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730

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CANT1	n.t.b					Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719
CASR	n.t.b					Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CBFB	n.t.b					Cleidocranial dysplasia 2, 620099
CBL	n.t.b					Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CC2D2A	n.t.b					COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCDC134	n.t.b					Osteogenesis imperfecta, type XXII, 619795
CCDC8	n.t.b					3-M syndrome 3, 614205
CCN2	n.t.b					?Spondyloepimetaphyseal dysplasia, Li-Shao-Li type, 621099;Kyphomelic dysplasia, 211350
CCN6	n.t.b					Progressive pseudorheumatoid dysplasia, 208230

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CCNQ	n.t.b					STAR syndrome, 300707
CDC42	n.t.b					Takenouchi-Kosaki syndrome, 616737
CDC45	n.t.b					Meier-Gorlin syndrome 7, 617063
CDC6	n.t.b					Meier-Gorlin syndrome 5, 613805
CDC73	n.t.b					Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001
CDH3	n.t.b					Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDK10	n.t.b					Al Kaissi syndrome, 617694
CDKN1C	n.t.b					IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CDT1	n.t.b					Meier-Gorlin syndrome 4, 613804
CENPE	n.t.b					?Microcephaly 13, primary, autosomal recessive, 616051
CEP120	n.t.b					Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761

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CEP152	n.t.b					Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823
CEP290	n.t.b					Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP57	n.t.b					Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	n.t.b					?Seckel syndrome 6, 614728
CFAP410	n.t.b					Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CHST11	n.t.b					?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	n.t.b					Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	n.t.b					Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHSY1	n.t.b					Temtamy preaxial brachydactyly syndrome, 605282

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CILK1	n.t.b					Cranioectodermal dysplasia 6, 621337;{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924;Endocrine-cerebroosteodysplasia, 612651
CKAP2L	n.t.b					Filippi syndrome, 272440
CLCN5	n.t.b					Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease 1, 300009;Nephrolithiasis, type I, 310468
CLCN7	n.t.b					Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
COG1	n.t.b					Congenital disorder of glycosylation, type IIg, 611209
COG4	n.t.b					Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COL10A1	n.t.b					Metaphyseal chondrodysplasia, Schmid type, 156500

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COL11A1	n.t.b					Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932
COL11A2	n.t.b					Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL1A1	n.t.b					Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420

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COL1A2	n.t.b					Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210
COL27A1	n.t.b					Steel syndrome, 615155

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COL2A1	n.t.b					?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;Spondylometaphyseal dysplasia, Algerian type, 184253;SED congenita, 183900;Kniest dysplasia, 156550;Stickler syndrome, type I, nonsyndromic ocular, 609508;Osteoarthritis with mild chondrodysplasia, 604864;Stickler syndrome, type I, 108300;Platyspondylic skeletal dysplasia, Torrance type, 151210;Spondyloepiphyseal dysplasia, Stanescu type, 616583;Avascular necrosis of the femoral head, 608805;Legg-Calve-Perthes disease, 150600;Epiphyseal dysplasia, multiple, with myopia and deafness, 132450
COL9A1	n.t.b					Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	n.t.b					Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284

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COL9A3	n.t.b					{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
COLEC10	n.t.b					3MC syndrome 3, 248340
COLEC11	n.t.b					3MC syndrome 2, 265050
COMP	n.t.b					Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
CPAP	n.t.b					Microcephaly 6, primary, autosomal recessive, 608393;Seckel syndrome 4, 613676
CPLANE1	n.t.b					Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CREB3L1	n.t.b					Osteogenesis imperfecta, type XVI, 616229
CREBBP	n.t.b					Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CRIPT	n.t.b					Rothmund-Thomson syndrome, type 3, 615789
CRTAP	n.t.b					Osteogenesis imperfecta, type VII, 610682

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CSF1R	n.t.b					Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSGALNACT1	n.t.b					Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CSPP1	n.t.b					Joubert syndrome 21, 615636
CTSA	n.t.b					Galactosialidosis, 256540;Brain small vessel disease 6 with leukoencephalopathy, 621394
CTSK	n.t.b					Pycnodysostosis, 265800
CUL7	n.t.b					3-M syndrome 1, 273750
CYP26B1	n.t.b					Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	n.t.b					Vitamin D-dependent rickets, type I, 264700
CYP2R1	n.t.b					Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
DDR1	n.t.b					
DDR2	n.t.b					Warburg-Cinotti syndrome, 618175;Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	n.t.b					Spondyloepimetaphyseal dysplasia, Shohat type, 602557

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DHCR24	n.t.b					Desmosterolosis, 602398
DHODH	n.t.b					Miller syndrome, 263750
DLL3	n.t.b					Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	n.t.b					Adams-Oliver syndrome 6, 616589
DLX3	n.t.b					Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510
DLX5	n.t.b					Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	n.t.b					
DMP1	n.t.b					Hypophosphatemic rickets, AR, 241520
DNA2	n.t.b					Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807
DNAJC21	n.t.b					Bone marrow failure syndrome 3, 617052
DNMT3A	n.t.b					Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
DOCK6	n.t.b					Adams-Oliver syndrome 2, 614219

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DONSON	n.t.b					Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230;Meier-Gorlin syndrome 10, 621528
DPCD	n.t.b					
DPF2	n.t.b					Coffin-Siris syndrome 7, 618027
DPM1	n.t.b					Congenital disorder of glycosylation, type Ie, 608799
DSE	n.t.b					Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DVL1	n.t.b					Robinow syndrome, autosomal dominant 2, 616331
DVL3	n.t.b					Robinow syndrome, autosomal dominant 3, 616894
DYM	n.t.b					Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800
DYNC2H1	n.t.b					Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	n.t.b					Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DYNC2I2	n.t.b					Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	n.t.b					Short-rib thoracic dysplasia 15 with polydactyly, 617088

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DYNLT2B	n.t.b					Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EBP	n.t.b					MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECEL1	n.t.b					Arthrogryposis, distal, type 5D, 615065
EDN1	n.t.b					Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706
EDNRA	n.t.b					{Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367
EFL1	n.t.b					Shwachman-Diamond syndrome 2, 617941
EFNB1	n.t.b					Craniofrontonasal dysplasia, 304110
EFTUD2	n.t.b					Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2AK3	n.t.b					Wolcott-Rallison syndrome, 226980
EIF4A3	n.t.b					Robin sequence with cleft mandible and limb anomalies, 268305
ELMO2	n.t.b					Vascular malformation, primary intraosseous, 606893
EN1	n.t.b					?ENDOVE syndrome, limb-brain type, 619218

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ENPP1	n.t.b					{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522
EOGT	n.t.b					Adams-Oliver syndrome 4, 615297
EP300	n.t.b					Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684
ERCC4	n.t.b					Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERF	n.t.b					Craniosynostosis 4, 600775;Chitayat syndrome, 617180
ERI1	n.t.b					Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663
ESCO2	n.t.b					Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300

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EVC	n.t.b					Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530
EVC2	n.t.b					Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530
EXOC6B	n.t.b					Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC5	n.t.b					Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXT1	n.t.b					Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300
EXT2	n.t.b					Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701
EXTL3	n.t.b					Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	n.t.b					Weaver syndrome, 277590
FAM111A	n.t.b					Kenny-Caffey syndrome, type 2, 127000;Gracile bone dysplasia, 602361
FAM20B	n.t.b					
FAM20C	n.t.b					Raine syndrome, 259775
FAM210A	n.t.b					

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FANCA	n.t.b					Fanconi anemia, complementation group A, 227650
FANCB	n.t.b					Fanconi anemia, complementation group B, 300514
FANCC	n.t.b					Fanconi anemia, complementation group C, 227645
FANCD2	n.t.b					Fanconi anemia, complementation group D2, 227646
FANCE	n.t.b					Fanconi anemia, complementation group E, 600901
FANCF	n.t.b					Fanconi anemia, complementation group F, 603467
FANCG	n.t.b					Fanconi anemia, complementation group G, 614082
FANCI	n.t.b					Fanconi anemia, complementation group I, 609053
FANCL	n.t.b					Fanconi anemia, complementation group L, 614083
FAR1	n.t.b					Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FBLN1	n.t.b					

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FBN1	n.t.b					Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900
FBN2	n.t.b					Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBXW4	n.t.b					
FERMT3	n.t.b					Leukocyte adhesion deficiency, type III, 612840
FGD1	n.t.b					Aarskog-Scott syndrome, 305400
FGF10	n.t.b					LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920
FGF16	n.t.b					Metacarpal 4-5 fusion, 309630
FGF23	n.t.b					Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100
FGF8	n.t.b					Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	n.t.b					Multiple synostoses syndrome 3, 612961

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FGFR1	n.t.b					Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	n.t.b					Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400

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FGFR3	n.t.b					Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
FIG4	n.t.b					Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FKBP10	n.t.b					Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450
FKBP14	n.t.b					Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557

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FLNA	n.t.b					Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620
FLNB	n.t.b					Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310
FMN1	n.t.b					
FN1	n.t.b					Spondylometaphyseal dysplasia, corner fracture type, 184255;Glomerulopathy with fibronectin deposits 2, 601894
FUCA1	n.t.b					Fucosidosis, 230000
FUZ	n.t.b					{Neural tube defects, susceptibility to}, 182940
FZD2	n.t.b					Omodysplasia 2, 164745
GALNS	n.t.b					Mucopolysaccharidosis IVA, 253000

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GALNT2	n.t.b					Congenital disorder of glycosylation, type IIt, 618885
GALNT3	n.t.b					Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GBA1	n.t.b					{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GCM2	n.t.b					Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343
GDF3	n.t.b					Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704

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GDF5	n.t.b					Acromesomelic dysplasia 2A, 200700;Acromesomelic dysplasia 2B, 228900;Multiple synostoses syndrome 2, 610017;Symphalangism, proximal, 1B, 615298;Brachydactyly, type A2, 112600;?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072
GDF6	n.t.b					Microphthalmia with coloboma 6, digenic, 613703;Microphthalmia, isolated 4, 613094;Leber congenital amaurosis 17, 615360;Multiple synostoses syndrome 4, 617898;Klippel-Feil syndrome 1, autosomal dominant, 118100
GH1	n.t.b					Kowarski syndrome, 262650;Growth hormone deficiency, isolated, type II, 173100;Growth hormone deficiency, isolated, type IB, 612781;Growth hormone deficiency, isolated, type IA, 262400
GHR	n.t.b					Laron dwarfism, 262500;Increased responsiveness to growth hormone, 604271;Growth hormone insensitivity, partial, 604271;{Hypercholesterolemia, familial, modifier of}, 143890

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GHRHR	n.t.b					Growth hormone deficiency, isolated, type IV, 618157
GHSR	n.t.b					Growth hormone deficiency, isolated partial, 615925
GINS2	n.t.b					
GJA1	n.t.b					Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850
GLB1	n.t.b					GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLI1	n.t.b					Polydactyly, preaxial I, 174400;Polydactyly, postaxial, type A8, 618123
GLI2	n.t.b					Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829

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GLI3	n.t.b					Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GMNN	n.t.b					Meier-Gorlin syndrome 6, 616835
GNAI3	n.t.b					Auriculocondylar syndrome 1, 602483
GNAS	n.t.b					Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;McCune-Albright syndrome, somatic mosaic, 174800;Pseudohypoparathyroidism Ib, 603233;Pseudopseudohypoparathyroidism, 612463;ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080
GNPAT	n.t.b					Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	n.t.b					?Rhizomelic dysplasia, Ain-Naz type, 619598
GNPTAB	n.t.b					Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	n.t.b					Mucolipidosis III gamma, 252605

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GNS	n.t.b					Mucopolysaccharidosis type IIID, 252940
GORAB	n.t.b					Geroderma osteodysplasticum, 231070
GPC3	n.t.b					Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	n.t.b					Keipert syndrome, 301026
GPC6	n.t.b					Omodysplasia 1, 258315
GPR161	n.t.b					{Medulloblastoma predisposition syndrome}, 155255
GPX4	n.t.b					Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GSC	n.t.b					Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GUSB	n.t.b					Mucopolysaccharidosis VII, 253220
GZF1	n.t.b					Joint laxity, short stature, and myopia, 617662
H19	n.t.b					
HAAO	n.t.b					Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HAPLN1	n.t.b					
HDAC4	n.t.b					Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC8	n.t.b					Cornelia de Lange syndrome 5, 300882

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HES7	n.t.b					Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	n.t.b					Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	n.t.b					Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HHAT	n.t.b					Nivelon-Nivelon-Mabille syndrome, 600092
HMGA2	n.t.b					Silver-Russell syndrome 5, 618908
HOXA11	n.t.b					Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	n.t.b					Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305
HOXD13	n.t.b					Syndactyly, type V, 186300;Synpolydactyly 1, 186000;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200;?Brachydactyly-syndactyly syndrome, 610713
HPGD	n.t.b					?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100

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HRAS	n.t.b					Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040
HS2ST1	n.t.b					Neurofacioskeletal syndrome with or without renal agenesis, 619194
HSPA9	n.t.b					Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPG2	n.t.b					Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800
HYLS1	n.t.b					Hydrolethalus syndrome, 236680
IARS2	n.t.b					Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
ID4	n.t.b					
IDH1	n.t.b					{Glioma, susceptibility to, somatic}, 137800
IDH2	n.t.b					D-2-hydroxyglutaric aciduria 2, 613657

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IDS	n.t.b					Mucopolysaccharidosis II, 309900
IDUA	n.t.b					Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IFIH1	n.t.b					Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFITM5	n.t.b					Osteogenesis imperfecta, type V, 610967
IFT122	n.t.b					Cranioectodermal dysplasia 1, 218330
IFT140	n.t.b					{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	n.t.b					Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	n.t.b					?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866

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IFT52	n.t.b					Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	n.t.b					Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	n.t.b					Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	n.t.b					Insulin-like growth factor I deficiency, 608747
IGF1R	n.t.b					Insulin-like growth factor I, resistance to, 270450
IGF2	n.t.b					Silver-Russell syndrome 3, 616489
IGFALS	n.t.b					Acid-labile subunit, deficiency of, 615961
IGSF1	n.t.b					Hypothyroidism, central, and testicular enlargement, 300888
IHH	n.t.b					Acrocapitofemoral dysplasia, 607778;Brachydactyly, type A1, 112500
IKBKB	n.t.b					Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204
IKBKG	n.t.b					Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081

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IL1RN	n.t.b					Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852
IL2RG	n.t.b					Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL6ST	n.t.b					Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752;Stuve-Wiedemann syndrome 2, 619751;Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523;?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750
INPPL1	n.t.b					Opsismodysplasia, 258480
INTU	n.t.b					?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925

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JAG1	n.t.b					?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500
KAT6B	n.t.b					SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KCNJ2	n.t.b					Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KDELR2	n.t.b					Osteogenesis imperfecta, type XXI, 619131
KIAA0586	n.t.b					Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	n.t.b					?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA0825	n.t.b					Polydactyly, postaxial, type A10, 618498
KIF22	n.t.b					Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF24	n.t.b					
KIF5B	n.t.b					Kyphomelic dysplasia, Itai-Ikegawa type, 621644

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KIF7	n.t.b					Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
KL	n.t.b					?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KMT2A	n.t.b					Wiedemann-Steiner syndrome, 605130
KRAS	n.t.b					Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;RAS-associated autoimmune leukoproliferative disorder 2, somatic, 621582;Bladder cancer, somatic, 109800
KYNU	n.t.b					?Hydroxykynureninuria, 236800;Vertebral, cardiac, renal, and limb defects syndrome 2, 617661

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LAMA5	n.t.b					Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076
LBR	n.t.b					Pelger-Huet anomaly, 169400;?Reynolds syndrome, 613471;Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019;Greenberg skeletal dysplasia, 215140
LBX1	n.t.b					?Central hypoventilation syndrome, congenital, 3, 619483
LEMD3	n.t.b					Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700
LFNG	n.t.b					Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	n.t.b					Pituitary hormone deficiency, combined, 3, 221750
LHX4	n.t.b					Pituitary hormone deficiency, combined, 4, 262700
LIFR	n.t.b					Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMBR1	n.t.b					Syndactyly, type IV, 186200;Laurin-Sandrow syndrome, 135750;Acheiropody, 200500;Triphalangeal thumb-polysyndactyly syndrome, 190605

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LMNA	n.t.b					Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112
LMX1B	n.t.b					Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LONP1	n.t.b					CODAS syndrome, 600373
LPIN2	n.t.b					Majeed syndrome, 609628
LRP4	n.t.b					?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780

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LRP5	n.t.b					Osteopetrosis, autosomal dominant 1, 607634;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;[Bone mineral density variability 1, high bone mass], 601884;Exudative vitreoretinopathy 4, 601813
LRRK1	n.t.b					Osteosclerotic metaphyseal dysplasia, 615198
LTBP1	n.t.b					Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	n.t.b					Glaucoma 3, primary congenital, D, 613086;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;?Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	n.t.b					Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
LYSET	n.t.b					Dysostosis multiplex, Ain-Naz type, 619345
LZTR1	n.t.b					Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670

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MAB21L2	n.t.b					Microphthalmia/colobo ma and skeletal dysplasia syndrome, 615877
MAD2L2	n.t.b					?Fanconi anemia, complementation group V, 617243
MAFB	n.t.b					Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300
MAN2B1	n.t.b					Mannosidosis, alpha-, types I and II, 248500
MANBA	n.t.b					Mannosidosis, beta, 248510
MAP2K1	n.t.b					Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis , isolated, somatic mosaic, 155950
MAP2K2	n.t.b					Cardiofaciocutaneous syndrome 4, 615280
MAP3K20	n.t.b					Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	n.t.b					Frontometaphyseal dysplasia 2, 617137;Cardiospondylo carpofacial syndrome, 157800
MAPK1	n.t.b					Noonan syndrome 13, 619087
MAPKAPK5	n.t.b					Neurocardiofaciodigital syndrome, 619869
MASP1	n.t.b					3MC syndrome 1, 257920

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MATN3	n.t.b					{Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728;Epiphyseal dysplasia, multiple, 5, 607078
MBTPS1	n.t.b					Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392;CAOP syndrome, 621252
MBTPS2	n.t.b					Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918
MCM5	n.t.b					?Meier-Gorlin syndrome 8, 617564
MECOM	n.t.b					Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MEGF8	n.t.b					Carpenter syndrome 2, 614976
MEOX1	n.t.b					Klippel-Feil syndrome 2, 214300
MESD	n.t.b					Osteogenesis imperfecta, type XX, 618644
MESP2	n.t.b					Spondylocostal dysostosis 2, autosomal recessive, 608681

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MET	n.t.b					Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705
MGP	n.t.b					Keutel syndrome, 245150
MIR140	n.t.b					Spondyloepiphyseal dysplasia, Nishimura type, 618618
MIR17HG	n.t.b					
MKS1	n.t.b					Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MMP13	n.t.b					?Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111;Metaphyseal dysplasia, Spahr type, 250400
MMP14	n.t.b					Winchester syndrome, 277950
MMP2	n.t.b					Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	n.t.b					Metaphyseal anadysplasia 2, 613073
MNX1	n.t.b					Currarino syndrome, 176450
MRAS	n.t.b					Noonan syndrome 11, 618499

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MSX2	n.t.b					Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500
MTAP	n.t.b					Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTX2	n.t.b					Mandibuloacral dysplasia progeroid syndrome, 619127
MYCN	n.t.b					Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748
MYH3	n.t.b					Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469;Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436;Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYL11	n.t.b					Arthrogryposis, distal, type 1C, 619110
MYO18B	n.t.b					Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
NADSYN1	n.t.b					Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGLU	n.t.b					?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920

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NANS	n.t.b					Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NBAS	n.t.b					Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483;?Acrofrontofacionasal dysostosis 3, 621629
NEK1	n.t.b					Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK9	n.t.b					?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022
NEPRO	n.t.b					
NEU1	n.t.b					Sialidosis, type II, 256550;Sialidosis, type I, 256550
NF1	n.t.b					Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFIX	n.t.b					Marshall-Smith syndrome, 602535;Malan syndrome, 614753

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NIN	n.t.b					?Seckel syndrome 7, 614851
NIPBL	n.t.b					Cornelia de Lange syndrome 1, 122470
NKX3-2	n.t.b					Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP3	n.t.b					CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendotheliti fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900
NMNAT1	n.t.b					Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553
NOG	n.t.b					Symphalangism, proximal, 1A, 185800;Brachydactyly, type B2, 611377;Stapes ankylosis with broad thumbs and toes, 184460;Tarsal-carpal coalition syndrome, 186570;Multiple synostoses syndrome 1, 186500
NOTCH1	n.t.b					Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	n.t.b					Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500

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NPPC	n.t.b					
NPR2	n.t.b					Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	n.t.b					Boudin-Mortier syndrome, 619543
NRAS	n.t.b					Noonan syndrome 6, 613224;RAS-associated autoimmune lymphoproliferative syndrome type 1, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
NSD1	n.t.b					Sotos syndrome, 117550
NSD2	n.t.b					Rauch-Steindl syndrome, 619695
NSDHL	n.t.b					CK syndrome, 300831;CHILD syndrome, 308050
NSMCE2	n.t.b					Seckel syndrome 10, 617253
NXN	n.t.b					Robinow syndrome, autosomal recessive 2, 618529

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OBSL1	n.t.b					3-M syndrome 2, 612921
OFD1	n.t.b					Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
ORC1	n.t.b					Meier-Gorlin syndrome 1, 224690
ORC4	n.t.b					Meier-Gorlin syndrome 2, 613800
ORC6	n.t.b					Meier-Gorlin syndrome 3, 613803
OSTM1	n.t.b					Osteopetrosis, autosomal recessive 5, 259720
OTX2	n.t.b					Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
P3H1	n.t.b					Osteogenesis imperfecta, type VIII, 610915
P4HB	n.t.b					Cole-Carpenter syndrome 1, 112240
PAM16	n.t.b					Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	n.t.b					Short stature, Dauber-Argente type, 619489
PAPSS2	n.t.b					Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847

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PAX3	n.t.b					Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PCNT	n.t.b					Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	n.t.b					Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680
PDE3A	n.t.b					Hypertension and brachydactyly syndrome, 112410
PDE4D	n.t.b					Acrodysostosis 2, with or without hormone resistance, 614613
PDGFRB	n.t.b					Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007
PEX5	n.t.b					Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716

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PEX6	n.t.b					Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	n.t.b					Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PHEX	n.t.b					Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	n.t.b					Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PIGV	n.t.b					Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PIK3R1	n.t.b					Immunodeficiency 36, 616005;SHORT syndrome, 269880;Agammaglobulinemia 7, autosomal recessive, 615214
PISD	n.t.b					Liberfarb syndrome, 618889
PITX1	n.t.b					Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	n.t.b					Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600

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PKDCC	n.t.b					Rhizomelic limb shortening with dysmorphic features, 618821
PLAG1	n.t.b					Adenomas, salivary gland pleomorphic, somatic, 181030;Silver-Russell syndrome 4, 618907
PLCB3	n.t.b					Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	n.t.b					Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
PLEKHM1	n.t.b					?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107
PLK4	n.t.b					Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	n.t.b					Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	n.t.b					Bruck syndrome 2, 609220
PLOD3	n.t.b					BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLS3	n.t.b					Bone mineral density QTL18, osteoporosis, 300910;Diaphragmatic hernia 5, X-linked, 306950

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PNPLA6	n.t.b					Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
POC1A	n.t.b					Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POLE	n.t.b					{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
POLL	n.t.b					
POLR1A	n.t.b					Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462
POLR1B	n.t.b					Treacher-Collins syndrome 4, 618939
POLR1C	n.t.b					Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	n.t.b					Treacher Collins syndrome 2, 613717
POLR3A	n.t.b					Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694

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POLR3B	n.t.b					Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3GL	n.t.b					Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POP1	n.t.b					Anauxetic dysplasia 2, 617396
POR	n.t.b					Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	n.t.b					Focal dermal hypoplasia, 305600
POU1F1	n.t.b					Pituitary hormone deficiency, combined or isolated, 1, 613038
PPIB	n.t.b					Osteogenesis imperfecta, type IX, 259440
PPM1D	n.t.b					Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450
PPP1CB	n.t.b					Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R21	n.t.b					Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383

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PRG4	n.t.b					Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRKACA	n.t.b					Cushing syndrome, ACTH-independent adrenal, somatic, 615830;Cardioacrofacial dysplasia 1, 619142
PRKACB	n.t.b					Cardioacrofacial dysplasia 2, 619143
PRKAR1A	n.t.b					Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960
PRKG2	n.t.b					Acromesomelic dysplasia 4, 619636;?Spondylometaphyseal dysplasia, Pagnamenta type, 619638
PRMT7	n.t.b					Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PROKR2	n.t.b					Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	n.t.b					Pituitary hormone deficiency, combined, 2, 262600
PSAT1	n.t.b					Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992

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PSMB1	n.t.b					?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038
PTBP1	n.t.b					STAD syndrome, 621495
PTDSS1	n.t.b					Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	n.t.b					Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045
PTHLH	n.t.b					Brachydactyly, type E2, 613382
PTPN11	n.t.b					Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PUF60	n.t.b					Verheij syndrome, 615583
PYCR1	n.t.b					Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	n.t.b					Carpenter syndrome 1, 201000
RAB33B	n.t.b					Smith-McCort dysplasia 2, 615222

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RAC3	n.t.b					Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	n.t.b					Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RAD51	n.t.b					Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244
RAF1	n.t.b					Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RALA	n.t.b					Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RASGRP2	n.t.b					Bleeding disorder, platelet-type, 18, 615888
RBBP8	n.t.b					Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic
RBM8A	n.t.b					Thrombocytopenia-absent radius syndrome, 274000
RBPJ	n.t.b					Adams-Oliver syndrome 3, 614814
RECQL4	n.t.b					Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280

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RFWD3	n.t.b					?Fanconi anemia, complementation group W, 617784
RIGI	n.t.b					Singleton-Merten syndrome 2, 616298
RIPPLY2	n.t.b					Spondylocostal dysostosis 6, 616566
RIT1	n.t.b					Noonan syndrome 8, 615355
RMRP	n.t.b					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RNPC3	n.t.b					Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU4ATAC	n.t.b					Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROR2	n.t.b					Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive 1, 268310
RPGRIP1L	n.t.b					Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPL10	n.t.b					{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RPL13	n.t.b					Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728

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RRAS	n.t.b					
RRAS2	n.t.b					Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	n.t.b					
RSPO2	n.t.b					?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022;Tetraamelia syndrome 2, 618021
RSPRY1	n.t.b					Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	n.t.b					Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510;Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Cleidocranial dysplasia, 119600
SALL1	n.t.b					Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	n.t.b					?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323
SATB2	n.t.b					Glass syndrome, 612313
SBDS	n.t.b					{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SCARF2	n.t.b					Van den Ende-Gupta syndrome, 600920

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SCUBE3	n.t.b					Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 2, 619184
SEC24D	n.t.b					Cole-Carpenter syndrome 2, 616294
SEMA3A	n.t.b					{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SERPINF1	n.t.b					Osteogenesis imperfecta, type VI, 613982
SERPINH1	n.t.b					{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848
SETD2	n.t.b					Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SF3B4	n.t.b					Acrofacial dysostosis 1, Nager type, 154400
SFRP4	n.t.b					Pyle disease, 265900
SGMS2	n.t.b					Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGSH	n.t.b					Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3BP2	n.t.b					Cherubism, 118400
SH3PXD2B	n.t.b					Frank-ter Haar syndrome, 249420

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SHH	n.t.b					Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SHOC2	n.t.b					Noonan syndrome-like with loose anagen hair 1, 607721
SHOX	n.t.b					Short stature, idiopathic familial, 300582;Leri-Weill dyschondrosteosis, 127300;Langer mesomelic dysplasia, 249700;Short stature, idiopathic familial, 300582;Langer mesomelic dysplasia, 249700;Leri-Weill dyschondrosteosis, 127300
SKI	n.t.b					Shprintzen-Goldberg syndrome, 182212
SLC10A7	n.t.b					Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC13A1	n.t.b					Hyposulfatemia with skeletal dysplasia, 621654
SLC17A5	n.t.b					Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC25A24	n.t.b					Fontaine progeroid syndrome, 612289

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SLC26A2	n.t.b					Epiphyseal dysplasia, multiple, 4, 226900;Diastrophic dysplasia, 222600;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Achondrogenesis IB, 600972;Atelosteogenesis, type II, 256050
SLC29A3	n.t.b					Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	n.t.b					Hypophosphatemic rickets with hypercalciuria, 241530
SLC35C1	n.t.b					Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	n.t.b					Schneckenbecken dysplasia, 269250
SLC39A13	n.t.b					Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	n.t.b					?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC4A2	n.t.b					?Osteopetrosis, autosomal recessive 9, 620366
SLCO2A1	n.t.b					Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441
SLCO5A1	n.t.b					

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SLX4	n.t.b					Fanconi anemia, complementation group P, 613951
SMAD2	n.t.b					Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	n.t.b					Loeys-Dietz syndrome 3, 613795
SMAD4	n.t.b					Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	n.t.b					Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
SMARCA4	n.t.b					Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMARCA5	n.t.b					
SMARCAL1	n.t.b					Schimke immunoosseous dysplasia, 242900
SMARCB1	n.t.b					Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322

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SMARCE1	n.t.b					{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SMC1A	n.t.b					Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	n.t.b					Cornelia de Lange syndrome 3, 610759
SMO	n.t.b					Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	n.t.b					Dentin dysplasia, type IA, 125400
SNRPB	n.t.b					Cerebrocostomandibular syndrome, 117650
SNX10	n.t.b					Osteopetrosis, autosomal recessive 8, 615085
SOS1	n.t.b					Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	n.t.b					Noonan syndrome 9, 616559
SOST	n.t.b					Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX2	n.t.b					Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900

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SOX3	n.t.b					Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX9	n.t.b					Campomelic dysplasia with autosomal sex reversal, 114290;46XY sex reversal 10, 616425;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290;46XX sex reversal 2, 278850
SP7	n.t.b					Osteogenesis imperfecta, type XII, 613849
SPARC	n.t.b					Osteogenesis imperfecta, type XVII, 616507
SPECC1L	n.t.b					Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251
SPINK5	n.t.b					Netherton syndrome, 256500
SPR	n.t.b					Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	n.t.b					Legius syndrome, 611431
SPRED2	n.t.b					Noonan syndrome 14, 619745
SRCAP	n.t.b					Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140

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SRP54	n.t.b					Neutropenia, severe congenital, 8, autosomal dominant, 618752
STAT3	n.t.b					Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	n.t.b					Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985
STIM1	n.t.b					Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
SULF1	n.t.b					
SUMF1	n.t.b					Multiple sulfatase deficiency, 272200
TAB2	n.t.b					Congenital heart defects, nonsyndromic, 2, 614980
TAPT1	n.t.b					Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897

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TBCE	n.t.b					Kenny-Caffey syndrome, type 1, 244460;Myopathy with rigid spine, scoliosis, and male-limited hypogonadotropic hypogonadism, 621734;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBX15	n.t.b					Cousin syndrome, 260660
TBX2	n.t.b					Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX3	n.t.b					Ulnar-mammary syndrome, 181450
TBX4	n.t.b					Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891;Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX5	n.t.b					Holt-Oram syndrome, 142900
TBX6	n.t.b					Spondylocostal dysostosis 5, 122600
TBXAS1	n.t.b					Ghosal hematodiaphyseal syndrome, 231095
TCF12	n.t.b					Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCIRG1	n.t.b					Osteopetrosis, autosomal recessive 1, 259700

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TCOF1	n.t.b					Treacher Collins syndrome 1, 154500
TCTN2	n.t.b					Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	n.t.b					Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TENT5A	n.t.b					Osteogenesis imperfecta, type XVIII, 617952
TGDS	n.t.b					Catel-Manzke syndrome, 616145
TGFB1	n.t.b					Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213;{Cystic fibrosis lung disease, modifier of}, 219700;Camurati-Engelmann disease 1, 131300
TGFB2	n.t.b					Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631
TGFB3	n.t.b					Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	n.t.b					{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	n.t.b					Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239

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THPO	n.t.b					Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
TMCO1	n.t.b					Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMEM165	n.t.b					Congenital disorder of glycosylation, type IIk, 614727
TMEM216	n.t.b					Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM231	n.t.b					Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM38B	n.t.b					Osteogenesis imperfecta, type XIV, 615066
TMEM53	n.t.b					Craniotubular dysplasia, Ikegawa type, 619727
TMEM67	n.t.b					Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TNFRSF11A	n.t.b					Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810

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TNFRSF11B	n.t.b					Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	n.t.b					Osteopetrosis, autosomal recessive 2, 259710
TONSL	n.t.b					Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TP63	n.t.b					Premature ovarian failure 21, 620311;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292;Hay-Wells syndrome, 106260;Split-hand/foot malformation 4, 605289;Orofacial cleft 8, 618149;Rapp-Hodgkin syndrome, 129400;ADULT syndrome, 103285;Limb-mammary syndrome, 603543
TRAF3IP1	n.t.b					
TRAF7	n.t.b					Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	n.t.b					Seckel syndrome 9, 616777
TRAPPC2	n.t.b					Spondyloepiphyseal dysplasia tarda, 313400
TREM2	n.t.b					{Alzheimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193

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TRIM37	n.t.b					Mulibrey nanism, 253250
TRIP11	n.t.b					Odontochondrodysplasia 1, 184260;Achondrogenesis, type IA, 200600
TRIP13	n.t.b					Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598
TRPS1	n.t.b					Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350
TRPV4	n.t.b					Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;SED, Maroteaux type, 184095;Metatropic dysplasia, 156530;Scapuloperoneal spinal muscular atrophy, 181405;Hereditary motor and sensory neuropathy, type IIc, 606071;?Avascular necrosis of femoral head, primary, 2, 617383;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500
TRPV6	n.t.b					Hyperparathyroidism, transient neonatal, 618188

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TTC21B	n.t.b					Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820
TTI2	n.t.b					Intellectual developmental disorder, autosomal recessive 39, 615541
TWIST1	n.t.b					Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TYROBP	n.t.b					Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
UBA2	n.t.b					ACCES syndrome, 619959
UBE2T	n.t.b					Fanconi anemia, complementation group T, 616435
UFSP2	n.t.b					?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028
VAC14	n.t.b					Striatonigral degeneration, childhood-onset, 617054
VDR	n.t.b					Rickets, vitamin D-resistant, type IIA, 277440

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VPS33A	n.t.b					Mucopolysaccharidosis-plus syndrome, 617303
VPS35L	n.t.b					Ritscher-Schinzel syndrome 3, 619135
WDR19	n.t.b					Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867
WDR35	n.t.b					Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WNT1	n.t.b					{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220
WNT10B	n.t.b					Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300
WNT3	n.t.b					?Tetra-amelia syndrome 1, 273395
WNT5A	n.t.b					Robinow syndrome, autosomal dominant 1, 180700
WNT6	n.t.b					
WNT7A	n.t.b					Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820;?Santos syndrome, 613005

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XRCC4	n.t.b					Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	n.t.b					Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	n.t.b					{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822
ZBTB16	n.t.b					Leukemia, acute promyelocytic, PL2F/RARA type
ZC4H2	n.t.b					Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	n.t.b					Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZSWIM6	n.t.b					Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors