SHORT STATURE AND SKELETAL DYSPLASIA DG 2.16 (354 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
ABCC9	142,6	100.0%	99.7%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAN	121,3	94.6%	89.1%	?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	172,6	100.0%	99.6%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	80,5	100.0%	99.7%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVR1	136,9	100.0%	99.9%	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	122,8	100.0%	99.8%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	109,2	97.6%	92.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	115,9	99.0%	96.3%	Geleophysic dysplasia 1, 231050
AGA	142,7	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGPS	75,4	99.5%	97.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
ALG12	155,7	100.0%	99.9%	Congenital disorder of glycosylation, type Ig, 607143
ALG3	106,5	100.0%	99.9%	Congenital disorder of glycosylation, type Id, 601110
ALG9	113	100.0%	99.6%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	172,8	100.0%	99.7%	Alstrom syndrome, 203800
ALPL	154,8	100.0%	99.7%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
AMER1	98,2	99.8%	98.9%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	97,4	99.8%	98.9%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990

ANKH	111,6	100.0%	99.9%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	119,6	99.2%	97.1%	KBG syndrome, 148050
ANO5	131	99.6%	97.3%	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ARSB	109,4	99.9%	98.9%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	80,5	97.9%	89.2%	Chondrodysplasia punctata, X-linked recessive, 302950
B3GALT6	81,7	82.6%	77.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	121	99.6%	96.5%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	123,9	99.8%	98.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BMP1	152,7	100.0%	100.0%	Osteogenesis imperfecta, type XIII, 614856
BMP2	163,4	100.0%	100.0%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMPER	127,1	100.0%	99.5%	Diaphanospondylodysostosis, 608022
BMPR1B	139,4	100.0%	100.0%	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BRF1	109	99.8%	98.1%	Cerebellofaciodental syndrome, 616202
BTK	98	99.9%	99.0%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
C21orf2	NC	NC	NC	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C5orf42	NC	NC	NC	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	137,4	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730

CANT1	144,9	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CBL	126	97.3%	97.0%	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CCDC8	186,9	100.0%	100.0%	3-M syndrome 3, 614205
CDC42	90,2	97.6%	89.1%	Takenouchi-Kosaki syndrome, 616737
CDC45	138,9	99.6%	98.1%	Meier-Gorlin syndrome 7, 617063
CDC6	139,4	99.9%	99.8%	?Meier-Gorlin syndrome 5, 613805
CDKN1C	100,1	89.8%	81.7%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDT1	130,9	100.0%	99.9%	Meier-Gorlin syndrome 4, 613804
CEP120	131,7	100.0%	99.4%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CHST3	133,8	100.0%	100.0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CLCN5	104,3	99.7%	96.5%	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	146,7	99.8%	98.7%	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
COG1	108,4	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209
COL10A1	106,8	100.0%	99.9%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	96,6	97.9%	94.0%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	111,6	100.0%	99.4%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150

COL1A1	141	99.8%	98.4%	Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL1A2	93,3	98.5%	94.6%	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL27A1	142,2	99.9%	99.2%	Steel syndrome, 615155
COL2A1	112,2	100.0%	99.7%	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL9A1	132,3	100.0%	99.7%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	95,2	99.9%	98.8%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	107,8	99.6%	96.8%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	180,6	100.0%	100.0%	3MC syndrome 2, 265050

COMP	132	95.8%	92.8%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
CREB3L1	135,8	100.0%	99.8%	Osteogenesis imperfecta, type XVI, 616229
CREBBP	110,7	99.4%	97.0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRTAP	120,2	100.0%	99.1%	Osteogenesis imperfecta, type VII, 610682
CSF1R	113,3	99.9%	99.1%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSGALNACT1	153	100.0%	99.6%	No OMIM phenotype Skeletal dysplasia and joint laxity (Vodopiutz (2017) Hum Mutat 38,34) ?Hemi-facial palsy (Saigoh (2011) J Hum Genet 56,143) ?Neuropathy, hereditary motor and sensory (Saigoh (2011) J Hum Genet 56,143)
CTSA	132,9	100.0%	99.9%	Galactosialidosis, 256540
CTSK	86,3	100.0%	99.8%	Pycnodysostosis, 265800
CUL7	129,2	100.0%	99.8%	3-M syndrome 1, 273750
CYP26B1	168,7	100.0%	100.0%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	147,1	100.0%	99.7%	Vitamin D-dependent rickets, type I, 264700
DDR2	115,8	100.0%	99.3%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 Warburg-Cinotti syndrome, 618175
DHCR24	155,8	100.0%	99.9%	Desmosterolosis, 602398
DLL3	108,5	96.7%	92.5%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX3	146,7	100.0%	99.0%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DMP1	133	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
DNMT3A	122,9	99.7%	98.2%	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DONSON	90,2	99.0%	92.4%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPM1	134,7	95.2%	88.2%	Congenital disorder of glycosylation, type Ie, 608799
DVL1	140,8	98.6%	95.9%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	186	100.0%	100.0%	Robinow syndrome, autosomal dominant 3, 616894
DYM	103,3	97.4%	95.5%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC2H1	102,2	98.8%	95.5%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95,4	99.7%	97.0%	Short-rib thoracic dysplasia 15 with polydactyly, 617088

EBP	63,2	99.5%	95.2%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EFL1	150,5	99.5%	98.1%	Shwachman-Diamond syndrome 2, 617941
EIF2AK3	134,2	99.5%	96.3%	Wolcott-Rallison syndrome, 226980
ENPP1	129,2	97.5%	93.3%	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
ESCO2	115,6	99.4%	97.3%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
EVC	106,3	95.9%	92.4%	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	110,2	99.4%	96.3%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXT1	88,6	99.6%	98.0%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	118	99.9%	99.1%	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	184,1	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FAM111A	232,1	100.0%	99.5%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM20C	145,2	100.0%	100.0%	Raine syndrome, 259775
FBN1	137,1	100.0%	99.8%	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FERMT3	144,9	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FGD1	86,7	98.4%	93.0%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400

FGF23	122,3	99.7%	97.7%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF8	130	97.9%	86.8%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	153,5	100.0%	100.0%	Multiple synostoses syndrome 3, 612961
FGFR1	122,6	100.0%	99.6%	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FGFR2	113,1	97.7%	96.8%	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	138,5	100.0%	99.6%	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300

				Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FIG4	157,5	100.0%	99.6%	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FKBP10	157,5	99.5%	97.3%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FLNA	142,7	100.0%	99.9%	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	123,6	99.7%	98.7%	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FN1	106	99.9%	98.9%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255
FUCA1	125,9	100.0%	99.9%	Fucosidosis, 230000
FZD2	179,4	99.7%	97.8%	Omodysplasia 2, 164745
GALNS	108,3	100.0%	99.3%	Mucopolysaccharidosis IVA, 253000
GALNT3	125,8	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GDF3	127,9	100.0%	100.0%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704

GDF5	169,6	100.0%	100.0%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 {Osteoarthritis-5}, 612400
GDF6	156,3	100.0%	100.0%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898
GH1	159,5	100.0%	100.0%	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650
GHR	150,6	99.5%	99.4%	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	108,1	96.0%	95.2%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	173,6	98.7%	95.2%	Growth hormone deficiency, isolated partial, 615925
GJA1	156,2	100.0%	100.0%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLI2	158,2	100.0%	100.0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829

GLI3	139,5	100.0%	99.3%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800
GMNN	124,4	99.7%	97.9%	Meier-Gorlin syndrome 6, 616835
GNAS	211,3	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNPAT	127,2	99.5%	96.8%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	148	100.0%	99.3%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	177,6	100.0%	98.5%	Mucolipidosis III gamma, 252605
GNS	94,5	99.6%	95.2%	Mucopolysaccharidosis type IIID, 252940
GORAB	165,7	100.0%	98.9%	Geroderma osteodysplasticum, 231070
GPC3	75,7	98.7%	92.7%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	126,6	100.0%	100.0%	Omodysplasia 1, 258315
GPR161	170,5	100.0%	100.0%	No OMIM phenotype Pituitary stalk interuption syndrome (Karaca (2015) J Clin Endocrinol Metab 100,E140)
GPX4	165,9	94.4%	90.7%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GUSB	99,5	92.5%	90.5%	Mucopolysaccharidosis VII, 253220
HDAC4	119,6	100.0%	99.9%	No OMIM phenotype Anorexia nervosa/bulimia nervosa (Cui (2013) J Clin Invest 123,4706) Brachydactyly mental retardation syndrome (Williams (2010) Am J Hum Genet 87, 219) ?Autism spectrum disorder (Pinto (2014) Am J Hum Genet 94, 677)
HES7	53,9	90.1%	72.7%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	66,2	100.0%	98.7%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230

HGSNAT	98,3	87.2%	86.2%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HMGA2	84,9	81.4%	75.5%	Leiomyoma, uterine, somatic, 150699
HOXA13	77,7	90.9%	79.6%	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HPGD	90,6	99.9%	98.9%	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HRAS	182,3	100.0%	100.0%	Bladder cancer, somatic, 109800 Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Thyroid carcinoma, follicular, somatic, 188470
HSPA9	82,6	89.5%	84.2%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPG2	119,8	99.5%	98.8%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	156,6	100.0%	100.0%	Hydrolethalus syndrome, 236680
IDH1	78	89.4%	77.3%	{Glioma, susceptibility to, somatic}, 137800
IDH2	98,5	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDS	100,9	99.9%	97.1%	Mucopolysaccharidosis II, 309900
IDUA	148,1	98.9%	94.6%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFITM5	94,9	100.0%	99.1%	Osteogenesis imperfecta, type V, 610967
IFT122	120,5	99.9%	99.0%	Cranioectodermal dysplasia 1, 218330
IFT140	117,6	99.9%	99.2%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	94,5	100.0%	99.4%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866

IFT80	64,9	96.7%	84.7%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGF1	98	100.0%	99.8%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	114,9	100.0%	99.6%	Insulin-like growth factor I, resistance to, 270450
IGF2	119,6	100.0%	100.0%	?Growth restriction, severe, with distinctive facies, 616489
IGFALS	108,2	100.0%	99.9%	Acid-labile subunit, deficiency of, 615961
IGSF1	69,9	99.2%	93.9%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	171,9	100.0%	100.0%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	110	99.3%	96.4%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	60,1	88.1%	78.8%	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL2RG	59,3	99.3%	94.0%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IMPAD1	170,4	100.0%	99.9%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INPPL1	127,9	99.8%	98.0%	Opsismodysplasia, 258480
KIAA0753	113,2	99.9%	98.7%	?Orofaciodigital syndrome XV, 617127
KIF22	173,8	100.0%	99.9%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	105,2	98.2%	93.5%	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KMT2A	133	100.0%	99.9%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130

KRAS	67,2	99.4%	97.3%	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LBR	103	98.3%	91.5%	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LEMD3	122,5	99.8%	98.4%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700
LFNG	117,6	92.8%	87.7%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	116,2	96.6%	96.4%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	131,2	100.0%	100.0%	Pituitary hormone deficiency, combined, 4, 262700
LIFR	110,3	99.7%	97.4%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMX1B	146,6	99.9%	98.5%	Nail-patella syndrome, 161200
LONP1	148	100.0%	100.0%	CODAS syndrome, 600373
LRP4	128	99.7%	99.0%	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	168,1	99.8%	98.7%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710

LRRK1	145,2	99.5%	97.9%	No OMIM phenotype Osteosclerotic metaphyseal dysplasia (Iida (2016) J Med Genet 53,568) ?Parkinson disease (Schulte (2013) Neurogenetics epub,epub)
LTBP2	112,9	99.9%	99.3%	?Weill-Marchesani syndrome 3, recessive, 614819 Glaucoma 3, primary congenital, D, 613086 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	147,5	100.0%	99.6%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	143,6	100.0%	99.7%	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MAN2B1	128,6	99.9%	98.6%	Mannosidosis, alpha-, types I and II, 248500
MANBA	118,3	99.5%	97.5%	Mannosidosis, beta, 248510
MAP2K1	92,3	99.5%	96.3%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	124,2	98.5%	94.1%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K7	118,4	99.9%	99.3%	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MATN3	99,6	86.5%	84.5%	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600
MEOX1	105	99.9%	97.4%	Klippel-Feil syndrome 2, 214300
MESP2	128	97.0%	94.9%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MGP	134,2	98.7%	94.6%	Keutel syndrome, 245150
MIR140	NC	NC	NC	No OMIM phenotype
MMP13	112,5	93.4%	92.1%	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	148	100.0%	99.7%	?Winchester syndrome, 277950
MMP2	154,2	100.0%	100.0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	143,9	100.0%	99.1%	Metaphyseal anadysplasia 2, 613073
MTAP	96	98.9%	93.4%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250

MYH3	94,1	99.9%	98.3%	Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Arthrogryposis, distal, type 2B (Sheldon-Hall), 601680 Arthrogryposis, distal, type 8, 178110
NAGLU	117,7	97.1%	94.1%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NANS	97,2	99.9%	98.4%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	138,5	99.9%	99.1%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEK1	115,9	99.7%	98.1%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK9	118,9	99.8%	98.2%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NEU1	141,3	99.3%	96.4%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NIN	127	99.9%	99.4%	?Seckel syndrome 7, 614851
NKX3-2	138,4	100.0%	99.8%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NOTCH2	123,7	100.0%	99.6%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPC	133	100.0%	100.0%	No OMIM phenotype
NPR2	144,1	100.0%	99.4%	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NRAS	145,5	100.0%	100.0%	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NXN	122,6	100.0%	100.0%	No OMIM phenotype
OBSL1	147,2	100.0%	99.8%	3-M syndrome 2, 612921

OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
ORC1	90,3	99.9%	98.7%	Meier-Gorlin syndrome 1, 224690
ORC4	73,6	98.1%	92.0%	Meier-Gorlin syndrome 2, 613800
ORC6	127,6	100.0%	99.9%	Meier-Gorlin syndrome 3, 613803
OSTM1	109,3	98.2%	92.5%	Osteopetrosis, autosomal recessive 5, 259720
OTX2	127,4	100.0%	99.3%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P3H1	129,3	100.0%	100.0%	Osteogenesis imperfecta, type VIII, 610915
P4HB	108,7	94.6%	93.8%	Cole-Carpenter syndrome 1, 112240
PAM16	64,5	66.4%	65.3%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	140,1	100.0%	99.7%	No OMIM phenotype Short stature (Dauber (2016) EMBO Mol Med epub,epub)
PAPSS2	103,8	99.7%	97.7%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PCNT	115,4	99.7%	97.7%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	95,6	97.9%	94.4%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE4D	102,7	95.8%	94.4%	Acrodysostosis 2, with or without hormone resistance, 614613
PEX5	107,9	100.0%	99.2%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	107,9	99.8%	98.6%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIK3R1	124,3	99.9%	98.9%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PISD	160,3	100.0%	99.9%	No OMIM phenotype
PITX1	174,5	98.9%	96.0%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550

PITX2	164,8	100.0%	99.5%	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PKDCC	86,8	94.7%	87.9%	No OMIM phenotype
PLEKHM1	127,8	100.0%	99.9%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLK4	149,7	99.8%	98.2%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD2	121,3	99.6%	97.3%	Bruck syndrome 2, 609220
PLS3	116,9	96.9%	95.3%	Bone mineral density QTL18, osteoporosis, 300910
POC1A	112,9	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POP1	120,1	100.0%	99.4%	Anauxetic dysplasia 2, 617396
POR	175,5	99.2%	97.1%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
POU1F1	109,1	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
PPIB	106,9	100.0%	100.0%	Osteogenesis imperfecta, type IX, 259440
PPP1CB	113,1	100.0%	99.1%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PRKAR1A	79,4	98.6%	92.6%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PROKR2	223	100.0%	100.0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	96,9	92.5%	83.7%	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	42,8	90.3%	72.5%	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PTDSS1	112	100.0%	99.9%	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	106,6	100.0%	99.1%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAB33B	191,3	100.0%	100.0%	Smith-McCort dysplasia 2, 615222
RAC3	121	98.3%	94.6%	No OMIM phenotype

RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	102,5	100.0%	99.7%	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	87,4	99.8%	97.4%	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	70,7	96.3%	87.0%	Adams-Oliver syndrome 3, 614814
RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RIPPLY2	78,7	100.0%	98.7%	?Spondylocostal dysostosis 6, 616566
RIT1	139,2	100.0%	100.0%	Noonan syndrome 8, 615355
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNPC3	41,6	88.8%	68.7%	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROR2	160,6	100.0%	99.7%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	123,4	96.7%	95.4%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	65,6	97.1%	86.9%	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RRAS	125,6	100.0%	99.1%	No OMIM phenotype
RSPRY1	142	100.0%	99.9%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	102,8	73.4%	72.2%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
SBDS	166,2	100.0%	100.0%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SCARF2	104,8	99.5%	95.7%	Van den Ende-Gupta syndrome, 600920
SEC24D	126,3	99.9%	99.5%	Cole-Carpenter syndrome 2, 616294
SERPINF1	104	100.0%	99.9%	Osteogenesis imperfecta, type VI, 613982

SERPINH1	195,8	100.0%	99.6%	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SGMS2	148,2	100.0%	100.0%	No OMIM phenotype
SGSH	140,2	97.6%	94.7%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	161,4	100.0%	99.9%	Frank-ter Haar syndrome, 249420
SHOC2	139,6	99.9%	99.4%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	35,9	82.5%	67.2%	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SLC10A7	111	99.9%	99.2%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC17A5	137,7	99.8%	96.1%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC25A24	128,9	99.6%	99.1%	Fontaine progeroid syndrome, 612289
SLC26A2	205,1	100.0%	99.9%	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC29A3	173,3	100.0%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	141,1	99.9%	99.0%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35D1	125	99.5%	97.2%	Schneckenbecken dysplasia, 269250
SLC39A13	145,1	100.0%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLCO2A1	97,7	99.9%	98.2%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	159,4	99.7%	98.3%	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95)
SMAD4	108,9	100.0%	99.9%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMARCAL1	113,2	100.0%	99.6%	Schimke immunoosseous dysplasia, 242900
SNRPB	77,7	99.9%	97.6%	Cerebrocostomandibular syndrome, 117650
SNX10	131,4	96.2%	95.7%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733

SOS2	99,7	99.7%	97.9%	Noonan syndrome 9, 616559
SOST	182,9	100.0%	99.6%	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX2	230	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	74	97.7%	92.9%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	159,9	100.0%	100.0%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP7	148,4	100.0%	99.3%	Osteogenesis imperfecta, type XII, 613849
SPARC	134,3	100.0%	100.0%	Osteogenesis imperfecta, type XVII, 616507
SPECC1L	127,5	100.0%	99.8%	?Facial clefting, oblique, 1, 600251 Hypertelorism, Teebi type, 145420 Opitz GBBB syndrome, type II, 145410
SPINK5	128	99.9%	99.5%	Netherton syndrome, 256500
SPR	145,7	100.0%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	146,5	99.8%	98.8%	Legius syndrome, 611431
SRCAP	153	100.0%	99.6%	Floating-Harbor syndrome, 136140
STAT3	103,2	100.0%	99.0%	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	114,1	99.8%	97.8%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
SULF1	135,6	99.9%	98.8%	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95) ?Hyperinsulinism (Proverbio (2013) PLoS One 8,e68740)
SUMF1	89,7	99.7%	96.8%	Multiple sulfatase deficiency, 272200
TAPT1	89,2	97.9%	92.2%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBCE	116,4	98.7%	94.7%	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBX15	106,3	100.0%	99.7%	Cousin syndrome, 260660
TBX4	175,9	99.5%	97.5%	Ischiocoxopodopatellar syndrome, 147891

TBX6	124,4	99.7%	96.5%	Spondylocostal dysostosis 5, 122600
TBXAS1	128,8	100.0%	100.0%	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	131,4	99.2%	96.6%	Osteopetrosis, autosomal recessive 1, 259700
TCTEX1D2	123,6	100.0%	99.4%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN2	122,4	99.9%	99.0%	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TGFB1	102,1	100.0%	99.6%	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 {Cystic fibrosis lung disease, modifier of}, 219700
TMEM165	148,2	100.0%	99.8%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	88	99.7%	95.7%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	101,1	100.0%	99.3%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM38B	107,9	100.0%	99.0%	Osteogenesis imperfecta, type XIV, 615066
TNFRSF11A	131	96.1%	95.2%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	172,4	100.0%	100.0%	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	129,8	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TONSL	125,7	100.0%	99.5%	No OMIM phenotype Pancreatic cancer (Smith (2015) Cancer Lett epub,epub) ?Schizophrenia (Fromer (2014) Nature 506, 179)
TRAPPC2	59,2	86.5%	67.4%	Spondyloepiphyseal dysplasia tarda, 313400
TRIP11	90,9	97.5%	92.6%	Achondrogenesis, type IA, 200600 Osteochondrodysplasia, 184260
TRPS1	154	100.0%	99.9%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351

TRPV4	138,4	100.0%	99.8%	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TTC21B	119,5	99.7%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
UFSP2	130,7	100.0%	99.5%	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
VDR	108,8	99.1%	96.0%	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR34	116,1	100.0%	100.0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	141,8	99.7%	98.4%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	108,1	99.7%	98.1%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WISP3	NC	NC	NC	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNT1	255,8	100.0%	99.8%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT5A	159	100.0%	100.0%	Robinow syndrome, autosomal dominant 1, 180700
XRCC4	143	99.9%	99.0%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	128,1	99.9%	98.2%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	147,5	99.7%	98.1%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800

ZBTB16	148,4	100.0%	100.0%	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZMPSTE24	128,7	100.0%	99.6%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors