SHORT STATURE AND SKELETAL DYSPLASIA GENE PANEL DG 2.14 (336 genes)

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCC9	157.9	99.9	99.2	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAN	121.6	91.6	85	?Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800
ACP5	196.2	100	99.9	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	129	99.1	94.2	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVR1	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	107.8	99.9	98.7	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	117.1	88.9	86.7	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	112.2	96.5	91	Geleophysic dysplasia 1, 231050
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AGPS	51.7	96.8	84.8	Rhizomelic chondrodysplasia punctata, type 3, 600121
ALG12	156.2	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG3	132.9	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG9	124.3	100	99.6	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
ALPL	156.4	100	100	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
AMER1	96.9	99.8	98.9	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	72.2	99	94	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990

ANKH	118.6	100	99.7	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	96.3	97.4	94.1	KBG syndrome, 148050
ANO5	142.2	99.5	95.9	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ARSB	117.5	94.9	87.7	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	102.1	99.2	93	Chondrodysplasia punctata, X-linked recessive, 302950
B3GALT6	47.5	76.4	71.7	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	93.6	99.4	95.9	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	104.3	96.1	95	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BMP1	143.9	99.9	99.1	Osteogenesis imperfecta, type XIII, 614856
BMP2	173.4	100	99.9	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMPER	159.9	99.9	99	Diaphanospondylodysostosis, 608022
BMPR1B	172.4	100	98.9	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BRAF	74.4	87.6	77.2	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	100	96.6	92.9	Cerebellofaciodental syndrome, 616202
BTK	116.2	100	99.6	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755
C21orf2	104.4	99.9	98.7	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
CA2	140.7	100	99.3	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730

CANT1	142.1	100	99.8	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CBL	129.8	96.9	95.7	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CCDC8	111.9	100	100	3-M syndrome 3, 614205
CDC42	97	96.7	89.5	Takenouchi-Kosaki syndrome, 616737
CDC45	160.7	99.4	97.5	Meier-Gorlin syndrome 7, 617063
CDC6	165.4	99.8	98.3	?Meier-Gorlin syndrome 5, 613805
CDKN1C	21.1	68.1	51.8	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDT1	99.3	96.8	93.8	Meier-Gorlin syndrome 4, 613804
CEP120	129.7	99.8	98.1	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CHST3	91.6	100	97.5	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CLCN5	134.6	99.6	98	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	129.7	99.5	98.2	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
COG1	124.2	100	99.9	Congenital disorder of glycosylation, type IIg, 611209
COL10A1	88.4	98.6	93.9	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	90.8	94.9	89.6	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	92.2	99.9	98.3	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL1A1	134.9	98.1	96.3	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	101.8	96.7	93.6	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 imperfecta, type III, 259420 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL2A1	103.4	99.9	99	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	121.2	99.5	96.9	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	65.1	98.3	88.8	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	65.5	95.6	86.7	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	203	100	100	3MC syndrome 2, 265050
COMP	121.4	93.6	92.4	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
CREB3L1	113.8	99.4	97	Osteogenesis imperfecta, type XVI, 616229

CREBBP	123.5	99.4	96.7	Rubinstein-Taybi syndrome 1, 180849
CRTAP	110.4	99.8	97.3	Osteogenesis imperfecta, type VII, 610682
CSGALNACT1	193.1	100	100	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	134.1	100	99.4	Galactosialidosis, 256540
CTSK	105	100	99.9	Pycnodysostosis, 265800
CUL7	149.6	99.8	97.9	3-M syndrome 1, 273750
CYP26B1	178.1	100	99.9	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	137.1	100	99.1	Vitamin D-dependent rickets, type I, 264700
DDR2	155	100	99.9	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DHCR24	183	100	100	Desmosterolosis, 602398
DLL3	64.1	88.8	79.9	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX3	109.8	100	99.1	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DMP1	159.5	99.9	99.1	Hypophosphatemic rickets, AR, 241520
DONSON	104.9	83.9	78.3	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPM1	131.1	91.7	86.7	Congenital disorder of glycosylation, type Ie, 608799
DVL1	113.9	97.8	94.1	Robinow syndrome, autosomal dominant 2, 616331
DYM	101.3	97.2	94.8	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC2H1	90.5	96.6	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EBP	83.3	100	98	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EFL1	174.8	99.4	97.7	Shwachman-Diamond syndrome 2, 617941
EIF2AK3	147.1	95.1	91.3	Wolcott-Rallison syndrome, 226980
ENPP1	134.8	92.4	83.2	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
EVC	110.4	93.2	89.8	?Weyers acrofacial dysostosis, 193530

				Ellis-van Creveld syndrome, 225500
EVC2	119.3	96.4	94.3	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXT1	105.4	99.9	98.5	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	163.5	99.9	99.1	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	206.4	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FAM111A	292.2	100	99.9	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM20C	101.3	100	98.9	Raine syndrome, 259775
FBN1	159.8	99.9	99.5	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	122.4	100	98.9	Leukocyte adhesion deficiency, type III, 612840
FGD1	85.7	92.7	86.5	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF23	106	99.9	97.8	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF8	111.4	90.2	79.7	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	165	100	100	Multiple synostoses syndrome 3, 612961
FGFR1	148	99.7	98.3	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	140.1	97.4	96.4	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	110.2	99.6	97	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	154.9	99.8	98.4	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FKBP10	158.6	96.9	92.8	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FLNA	138.1	100	99.5	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400

				Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	150	99.8	99.2	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FN1	145.4	100	99.5	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255
FUCA1	135	100	99.5	Fucosidosis, 230000
FZD2	176.6	98.3	94.8	No OMIM phenotype Omodysplasia, autosomal dominant (Saal (2015) Hum Mol Genet 24,3399)
GALNS	93.2	99	95.6	Mucopolysaccharidosis IVA, 253000
GALNT3	128.2	99.2	96	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GDF3	134.9	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	141.8	100	100	?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	75.2	98.7	89	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	175.2	100	100	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	212.3	99.8	99.5	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	116.4	95.3	94.7	Growth hormone deficiency, isolated, type IB, 612781
GHSR	206	99.9	98.4	Growth hormone deficiency, isolated partial, 615925
GJA1	246.4	100	100	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	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLI2	138.5	99.4	97.4	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	154.2	100	99.7	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	101.2	92.6	83.8	Meier-Gorlin syndrome 6, 616835
GNAS	141	98.5	95.9	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	133.6	99.4	96.4	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	151.6	96.1	89.7	Mucolipidosis III gamma, 252605
GNS	107.9	96.9	92	Mucopolysaccharidosis type IIID, 252940
GORAB	176.3	99.7	97.8	Geroderma osteodysplasticum, 231070
GPC6	142	100	100	Omodysplasia 1, 258315
GPR161	196.8	100	100	No OMIM phenotype Pituitary stalk interuption syndrome (Karaca (2015) J Clin Endocrinol Metab 100,E140)
GPX4	119.2	85.2	76.6	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GUSB	116.1	92.2	89.4	Mucopolysaccharidosis VII, 253220
HDAC4	111.9	99.9	99.3	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	29.1	64.9	42.6	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	57.6	99.2	92.6	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HGSNAT	101	86.4	85.7	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HMGA2	78.8	84	76.5	Leiomyoma, uterine, somatic, 150699
HOXA13	49	69.2	61.7	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HPGD	88	100	98.5	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HRAS	164.7	99.8	98.1	Congenital myopathy with excess of muscle spindles, 218040

				Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HSPA9	91.6	91.1	85.9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPG2	121.3	99.4	98.2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	171.1	100	100	Hydrolethalus syndrome, 236680
IDH1	97.9	92.5	81.7	{Glioma, susceptibility to, somatic}, 137800
IDH2	103.5	99.6	96.9	D-2-hydroxyglutaric aciduria 2, 613657
IDS	111.3	99.6	98.3	Mucopolysaccharidosis II, 309900
IDUA	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFITM5	63.4	99.4	94.9	Osteogenesis imperfecta, type V, 610967
IFT122	152	100	99.9	Cranioectodermal dysplasia 1, 218330
IFT140	114.7	99.9	99	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99.6	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	114.8	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT80	57.8	87.6	70.7	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGF1	122.5	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	144	100	99.8	Insulin-like growth factor I, resistance to, 270450
IGF2	100	100	100	?Growth restriction, severe, with distinctive facies, 616489
IGFALS	79.7	99.9	96.8	Acid-labile subunit, deficiency of, 615961
IGSF1	84.8	99.5	96.6	Hypothyroidism, central, and testicular enlargement, 300888
IHH	129.3	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500

IKBKB	123.5	98.5	94.2	Immunodeficiency 15, 615592
IKBKG	52.5	84.6	73.2	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 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	65.2	99.8	97.3	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IMPAD1	147.2	99.9	99.4	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INPPL1	123.6	96.7	93.7	Opsismodysplasia, 258480
KIAA0753	123.9	99.9	98.7	?Orofaciodigital syndrome XV, 617127
KIF22	163.1	100	99.9	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	85.7	93.5	88.9	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KMT2A	152.5	99.3	98.6	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KRAS	64.7	99.9	98.7	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	87.8	93.3	83.9	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LEMD3	96.7	95.4	88.8	Buschke-Ollendorff syndrome, 166700

				Osteopoikilosis with or without melorheostosis, 166700
LFNG	91.6	85.4	83.3	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	84.7	94	80.6	Pituitary hormone deficiency, combined, 3, 221750
LHX4	144.9	100	99.8	Pituitary hormone deficiency, combined, 4, 262700
LIFR	123.4	97.2	92.1	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMX1B	111.4	97	92.3	Nail-patella syndrome, 161200
LONP1	141.5	97.9	96.4	CODAS syndrome, 600373
LRP4	166.6	99.1	98.9	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	189.8	98.2	97.9	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	154.1	98.9	97.1	No OMIM phenotype Osteosclerotic metaphyseal dysplasia (Iida (2016) J Med Genet 53,568) ?Parkinson disease (Schulte (2013) Neurogenetics epub,epub)
LTBP2	104.6	99.6	97.1	?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	113.5	98.7	94.7	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	134	100	99.4	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MAN2B1	122.3	99.1	96.2	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99.7	97.2	Mannosidosis, beta, 248510
MAP2K1	92.3	99.8	95.6	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	107.9	97.6	89.2	Cardiofaciocutaneous syndrome 4, 615280

MAP3K7	114.6	99.7	98.1	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MATN3	116.6	84.7	84.7	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600
MEOX1	76.8	96.6	91.2	Klippel-Feil syndrome 2, 214300
MESP2	81.5	93.1	87.9	Spondylocostal dysostosis 2, autosomal recessive, 608681
MGP	132	92.7	91.6	Keutel syndrome, 245150
MMP13	124.1	93.5	91.5	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	152.1	100	99.5	?Winchester syndrome, 277950
MMP2	164.4	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	125.4	96.2	91.8	Metaphyseal anadysplasia 2, 613073
MTAP	109.2	93.6	86.5	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYH3	110.4	99.9	98.6	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
NAGLU	108.7	92.4	90.4	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NANS	106.1	100	99.9	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	145.3	99.5	97.6	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEK1	103.2	98.1	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK9	136.8	99.7	98.5	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NEU1	148.1	99.4	97.1	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NIN	143.5	99.7	98.6	?Seckel syndrome 7, 614851
NKX3-2	55.8	92.4	73.9	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NOTCH2	172.4	100	99.9	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500

NPPC	87.8	99.8	96.7	No OMIM phenotype
NPR2	164.8	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NRAS	188.4	100	100	?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	86	99.7	96.5	No OMIM phenotype ?Robinow syndrome (White (2018) Am J Hum Genet 102,27)
OBSL1	140.5	99.8	98.5	3-M syndrome 2, 612921
OFD1	51.5	84	67.8	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
ORC1	106.7	99.9	98.9	Meier-Gorlin syndrome 1, 224690
ORC4	57.6	95.8	82	Meier-Gorlin syndrome 2, 613800
ORC6	126.8	100	100	Meier-Gorlin syndrome 3, 613803
OSTM1	80.7	90.8	88.4	Osteopetrosis, autosomal recessive 5, 259720
OTX2	154.8	100	99.8	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P3H1	137	100	99.9	Osteogenesis imperfecta, type VIII, 610915
P4HB	105.1	94.6	94.4	Cole-Carpenter syndrome 1, 112240
PAM16	50.7	65.2	64.7	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	175.4	100	99.8	No OMIM phenotype Short stature (Dauber (2016) EMBO Mol Med epub,epub)
PAPSS2	108.5	99.7	98.5	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PCNT	117.6	98.9	96	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	113.5	98.3	94.7	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940

PDE4D	101.2	92.8	88.3	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PEX5	111.7	99.9	98.3	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	113.5	89.6	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	125	99.9	98	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	115.6	100	99.8	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIK3R1	129.3	99.7	97.3	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PITX1	144.6	91.4	86.8	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	147.8	99.7	97.5	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PLEKHM1	141.1	100	99.9	Osteopetrosis, autosomal recessive 6, 611497
PLK4	145.5	99.5	96.3	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD2	108.6	94.7	88.6	Bruck syndrome 2, 609220
PLS3	131.5	96.8	95.2	Bone mineral density QTL18, osteoporosis, 300910
POC1A	133.8	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POP1	114.2	100	99.7	Anauxetic dysplasia 2, 617396
POU1F1	106.3	98.2	94.7	Pituitary hormone deficiency, combined, 1, 613038
PPIB	118.4	100	100	Osteogenesis imperfecta, type IX, 259440
PPP1CB	96.6	99.6	98.4	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PRKAR1A	90.7	99.1	93.9	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	331.8	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	76.5	91.6	84.3	Pituitary hormone deficiency, combined, 2, 262600

PSAT1	53.2	91.4	75.8	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
PTDSS1	127.2	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	108.5	99.9	98.8	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAB33B	233.9	100	100	Smith-McCort dysplasia 2, 615222
RAF1	127.3	100	99.7	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	97.5	99.9	98.6	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	106.6	100	99.4	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	89.2	94.1	86.4	Adams-Oliver syndrome 3, 614814
RIPPLY2	63.5	99	83.8	?Spondylocostal dysostosis 6, 616566
RIT1	165.6	100	100	Noonan syndrome 8, 615355
RMRP				Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNPC3	41.3	85.1	61.1	No OMIM phenotype Growth hormone deficiency (Argente (2014) EMBO Mol Med epub, epub)
RNU4ATAC				Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROR2	165.9	99.4	98	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	126.2	96.4	93.9	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	85.9	99.1	93.1	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847

RRAS	116.3	89.4	81.6	No OMIM phenotype
RSPRY1	168.9	100	99.9	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	106.4	72.3	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	212.3	100	99.9	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SCARF2	82.5	85.2	73.6	Van den Ende-Gupta syndrome, 600920
SEC24D	136.9	99.9	98.6	Cole-Carpenter syndrome 2, 616294
SERPINF1	101.8	100	99.4	Osteogenesis imperfecta, type VI, 613982
SERPINH1	183.7	100	99.9	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SGSH	129	95.1	93.6	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	140.2	100	99.8	Frank-ter Haar syndrome, 249420
SHOC2	140.4	100	99.4	Noonan-like syndrome with loose anagen hair, 607721
SHOX	29.1	73.5	61.4	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SLC10A7	110	100	98.9	No OMIM phenotype
SLC17A5	119.6	96.8	92.9	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC25A24	115.8	98.6	96.3	Fontaine progeroid syndrome, 612289
SLC26A2	233.2	100	100	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	203.6	99.9	99.5	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	105.6	98.9	94.5	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35D1	115.4	95.7	90.4	Schneckenbecken dysplasia, 269250
SLC39A13	114.8	99.8	98	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLCO2A1	110.5	100	99.6	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441

SLCO5A1	169.1	99.8	97.7	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95)
SMAD4	125.5	100	100	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMARCAL1	134.6	100	99.9	Schimke immunoosseous dysplasia, 242900
SNRPB	75.2	99.8	97.4	Cerebrocostomandibular syndrome, 117650
SNX10	118.9	96.2	96.1	Osteopetrosis, autosomal recessive 8, 615085
SOS1	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	97.1	98.5	92.8	Noonan syndrome 9, 616559
SOST	112.6	100	99.6	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX2	128.8	98.3	93.1	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	37.7	86.4	71.5	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	134	97.8	93.8	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP7	159.1	99.9	99.3	?Osteogenesis imperfecta, type XII, 613849
SPARC	161	100	100	Osteogenesis imperfecta, type XVII, 616507
SPINK5	145	99.4	96.5	Netherton syndrome, 256500
SPR	166.5	98.9	90	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	164.3	98.7	96.7	Legius syndrome, 611431
SRCAP	153.9	99.8	99.1	Floating-Harbor syndrome, 136140
STAT3	119.5	99.9	99	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	130.6	99.7	97.2	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
SULF1	164.2	100	99.9	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95)

				?Hyperinsulinism (Proverbio (2013) PLoS One 8,e68740)
SUMF1	103.3	98.6	91.1	Multiple sulfatase deficiency, 272200
TAPT1	83.5	88.5	85.9	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBCE	128	99.9	98.2	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBX15	131.7	100	99.4	Cousin syndrome, 260660
TBX4	170.7	94.9	92.8	Ischiocoxopodopatellar syndrome, 147891
TBX6	122.1	91.5	79.7	Spondylocostal dysostosis 5, 122600
TBXAS1	140.3	100	100	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	113.5	95.4	89.4	Osteopetrosis, autosomal recessive 1, 259700
TCTEX1D2	126.1	100	99.3	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN2	144.2	99.5	97	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	127.6	100	99.8	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TGFB1	86.9	99.7	95.1	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TMEM165	113.9	99.8	98.1	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	111.9	100	98.7	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.5	100	99.9	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM38B	114.3	100	99.4	Osteogenesis imperfecta, type XIV, 615066
TNFRSF11A	146.3	93.3	91.4	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	224.8	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	150.4	99.3	93.2	Osteopetrosis, autosomal recessive 2, 259710
TRAPPC2	85.3	87	66.7	Spondyloepiphyseal dysplasia tarda, 313400
TRIP11	84.3	95.2	87.4	Achondrogenesis, type IA, 200600
TRPS1	175	100	99.8	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351

TRPV4	172.4	99.5	98.7	?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	100.7	99.7	97.6	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
VDR	123.3	98	95.2	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
WDR19	132.1	99.8	98.1	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR34	106.6	99.5	96.2	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	145.1	99.3	97.7	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	114.2	99.1	96.3	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WISP3	118.4	100	100	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNT1	188.8	100	99.9	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT5A	155.7	100	100	Robinow syndrome, autosomal dominant 1, 180700
XRCC4	103.2	99.7	97.3	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	132.5	90.4	87.1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	136.3	98.9	94.9	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
ZBTB16	151.4	100	100	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447

ZMPSTE24	113.3	100	99.1	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 : September 11th, 2018.
This list is accurate for panel version DG 2.14
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors