

SHORT STATURE AND SKELETAL DYSPLASIA GENE PANEL DG 2.12 (326 genes)

<i>Gene</i>	<i>Median</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
ABCC9	157.8	99	99	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 10, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAN	121.8	91	85	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACP5	196.7	100	99	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	129.3	99	94	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACVR1	165.2	100	100	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	108	99	98	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	117.3	88	86	Weill-Marchesani-like syndrome, 613195
ADAMTSL2	112.4	96	91	Geleophysic dysplasia 1, 231050
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AGPS	51.7	96	84	Rhizomelic chondrodysplasia punctata, type 3, 600121
ALG12	156.3	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG3	133	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG9	117.1	100	99	Congenital disorder of glycosylation, type II, 608776 Gillesen-Kaesbach-Nishimura syndrome, 263210
ALMS1	179.4	99	99	Alstrom syndrome, 203800
ALPL	156.7	100	100	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
AMER1	97.6	99	98	Osteopathia striata with cranial sclerosis, 300373
ANKH	118.7	100	99	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000

ANKRD11	96.4	97	94	KBG syndrome, 148050
ANO5	142.1	99	96	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ARSB	117.4	94	87	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	102.9	99	93	Chondrodysplasia punctata, X-linked recessive, 302950
B3GALT6	47.6	76	71	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	93.5	99	95	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	104.4	96	95	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BMP1	141.6	99	99	Osteogenesis imperfecta, type XIII, 614856
BMPER	159.9	99	98	Diaphanospondylodysostosis, 608022
BMPR1B	172.1	100	98	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BRAF	74.3	87	77	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706
BTK	115.5	100	99	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755
C21orf2	104.8	99	98	No OMIM phenotype Retinal dystrophy, early-onset with macular staphyloma (Khan (2015) Br J Ophthalmol 99,1725) Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236) Jeune syndrome (Wheway (2015) Nat Cell Biol 17,1074)
CA2	140.5	100	99	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CANT1	142.4	100	99	Desbuquois dysplasia 1, 251450
CBL	133.2	99	98	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CCDC8	112.1	100	100	3-M syndrome 3, 614205

CDC45	160.7	99	97	Meier-Gorlin syndrome 7, 617063
CDC6	165.5	99	98	?Meier-Gorlin syndrome 5, 613805
CDKN1C	21.3	68	51	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDT1	99.5	96	93	Meier-Gorlin syndrome 4, 613804
CEP120	128.1	99	98	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CHST3	91.8	100	97	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CLCN5	135.4	99	98	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	130	99	98	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
COG1	124	100	99	Congenital disorder of glycosylation, type IIg, 611209
COL10A1	88.5	98	93	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	90.8	94	89	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	13.2	14	14	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610
COL1A1	135.1	98	96	Caffey disease, 114000 Ehlers-Danlos syndrome, classic, 130000 Ehlers-Danlos syndrome, type VIIA, 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	101.9	96	93	Ehlers-Danlos syndrome, cardiac valvular form, 225320 Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL2A1	103.6	99	98	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 Otospondylomegaepiphyseal dysplasia, 215150 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler syndrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL9A1	120.5	99	96	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	65.1	98	88	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
COL9A3	65.4	95	86	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	203.1	100	100	3MC syndrome 2, 265050
COMP	121.8	93	92	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
CREB3L1	113.9	99	97	No OMIM phenotype Osteogenesis imperfecta, autosomal recessive (Symoens (2013) Orphanet J Rare Dis 8)
CREBBP	123.5	99	96	Rubinstein-Taybi syndrome, 180849

CRTAP	110.6	99	97	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	134.4	100	99	Galactosialidosis, 256540
CTSK	105.1	100	99	Pycnodysostosis, 265800
CUL7	149.9	99	97	3-M syndrome 1, 273750
CYP26B1	178.4	100	99	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	137.4	100	99	Vitamin D-dependent rickets, type I, 264700
DDR2	155.2	100	99	Spondylometaphyseal dysplasia, short limb-hand type, 271665
DHCR24	177.4	100	100	Desmosterolosis, 602398
DLL3	64.3	88	80	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX3	110.1	100	99	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DMP1	159.9	99	99	Hypophosphatemic rickets, AR, 241520
DONSON	104.9	83	78	No OMIM phenotype Microcephalic dwarfism (Reynolds (2017) Nat Genet 49,537)
DPM1	131.2	91	86	Congenital disorder of glycosylation, type Ie, 608799
DVL1	114.1	97	94	Robinow syndrome, autosomal dominant 2, 616331
DYM	101.2	97	94	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC2H1	90.4	96	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EBP	83.8	100	97	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EIF2AK3	147	95	91	Wolcott-Rallison syndrome, 226980
ENPP1	134.9	92	83	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	110.7	93	89	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	119	96	94	Ellis-van Creveld syndrome, 225500

				Weyers acrofacial dysostosis, 193530
EXT1	105.5	99	98	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	163.6	99	99	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
EXTL3	206.6	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FAM111A	292	100	99	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM20C	101.4	100	98	Raine syndrome, 259775
FBN1	159.8	99	99	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection 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	122.6	100	98	Leukocyte adhesion deficiency,type III,612840
FGD1	86.4	92	86	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF23	106.4	99	97	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF8	111.5	90	79	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	165.1	100	100	?Multiple synostoses syndrome 3, 612961
FGFR1	148.3	99	98	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	142.4	97	96	<p>Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410</p> <p>Apert syndrome, 101200</p> <p>Beare-Stevenson cutis gyrata syndrome, 123790</p> <p>Bent bone dysplasia syndrome, 614592</p> <p>Craniofacial-skeletal-dermatologic dysplasia, 101600</p> <p>Craniosynostosis, nonspecific</p> <p>Crouzon syndrome, 123500</p> <p>Gastric cancer, somatic, 613659</p> <p>Jackson-Weiss syndrome, 123150</p> <p>LADD syndrome, 149730</p> <p>Pfeiffer syndrome, 101600</p> <p>Saethre-Chotzen syndrome, 101400</p> <p>Scaphocephaly and Axenfeld-Rieger anomaly</p> <p>Scaphocephaly, maxillary retrusion, and mental retardation, 609579</p>
FGFR3	110.5	99	97	<p>Achondroplasia, 100800</p> <p>Bladder cancer, somatic, 109800</p> <p>CATSHL syndrome, 610474</p> <p>Cervical cancer, somatic, 603956</p> <p>Colorectal cancer, somatic, 114500</p> <p>Crouzon syndrome with acanthosis nigricans, 612247</p> <p>Hypochondroplasia, 146000</p> <p>LADD syndrome, 149730</p> <p>Muenke syndrome, 602849</p> <p>Nevus, epidermal, somatic, 162900</p> <p>SADDAN, 616482</p> <p>Spermatocytic seminoma, somatic, 273300</p> <p>Thanatophoric dysplasia, type I, 187600</p> <p>Thanatophoric dysplasia, type II, 187601</p>
FIG4	154.9	99	98	<p>Amyotrophic lateral sclerosis 11, 612577</p> <p>Charcot-Marie-Tooth disease, type 4J, 611228</p> <p>Yunis-Varon syndrome, 216340</p> <p>?Polymicrogyria, bilateral temporooccipital, 612691</p>
FKBP10	153.3	97	93	<p>Bruck syndrome 1, 259450</p> <p>Osteogenesis imperfecta, type XI, 610968</p>

FLNA	139.2	100	99	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	152.7	100	99	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylcarpotarsal synostosis syndrome, 272460
FN1	145.5	100	99	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FUCA1	134.9	100	99	Fucosidosis, 230000
FZD2	176.6	98	94	No OMIM phenotype Omodysplasia, autosomal dominant (Saal (2015) Hum Mol Genet 24,3399)
GALNS	93.1	99	95	Mucopolysaccharidosis IVA, 253000
GALNT3	128	99	96	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GDF3	135.1	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	141.9	100	100	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 ?Acromesomelic dysplasia, Hunter-Thompson type, 201250 {Osteoarthritis-5}, 612400

GDF6	75	98	89	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GH1	175.4	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	213.4	99	99	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	122.8	100	99	Growth hormone deficiency, isolated, type IB, 612781
GHSR	206.3	99	98	Growth hormone deficiency, isolated partial, 615925
GJA1	246.5	100	100	Atrioventricular septal defect 3, 600309 Cranio metaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratoderma variabilis et progressiva, 133200 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	91.6	99	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLI2	138.7	99	97	Culler-Jones syndrome, 615849 Holoprosencephaly-9, 610829
GLI3	154.4	100	99	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	83	Meier-Gorlin syndrome 6, 616835

GNAS	141.2	98	95	Acromegaly, somatic, 102200 ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNPAT	133.7	99	96	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	167.6	98	97	Mucopolipidosis II alpha/beta, 252500 Mucopolipidosis III alpha/beta, 252600
GNPTG	153.1	96	90	Mucopolipidosis III gamma, 252605
GNS	107.9	96	92	Mucopolysaccharidosis type IIID, 252940
GORAB	176.3	99	97	Geroderma osteodysplasticum, 231070
GPC6	142	100	100	Omodysplasia 1, 258315
GPR161	197	100	100	No OMIM phenotype Pituitary stalk interruption syndrome (Karaca (2015) J Clin Endocrinol Metab 100,E140)
GPX4	161.9	86	85	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GUSB	116.4	92	89	Mucopolysaccharidosis VII, 253220
HDAC4	113.3	99	99	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	29.2	65	42	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	57.8	99	92	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septo-optic dysplasia, 182230
HGSNAT	95.2	81	80	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HMGA2	78.8	84	76	Leiomyoma, uterine, somatic, 150699
HOXA13	49.3	69	61	Guttacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HPGD	87.9	100	98	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900

				Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HRAS	164.9	99	98	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	85	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPG2	121.4	99	98	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	171.2	100	100	Hydrolethalus syndrome, 236680
IDH1	97.9	92	81	{Glioma, susceptibility to, somatic}, 137800
IDH2	103.7	99	96	D-2-hydroxyglutaric aciduria 2, 613657
IDS	110.7	99	98	Mucopolysaccharidosis II, 309900
IDUA	121.3	89	82	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFITM5	63.3	99	94	Osteogenesis imperfecta, type V, 610967
IFT122	152.3	100	99	Cranioectodermal dysplasia 1, 218330
IFT140	115	100	99	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	114.8	100	100	Cranioectodermal dysplasia 3, 614099
IFT80	57.6	87	70	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGF1	122.7	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	144.2	100	99	Insulin-like growth factor I, resistance to, 270450
IGF2	100.2	100	100	?Growth restriction, severe, with distinctive facies, 616489
IGFALS	79.7	99	96	Acid-labile subunit, deficiency of, 615961
IGSF1	85.5	99	96	Hypothyroidism, central, and testicular enlargement, 300888
IHH	129.4	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500

IKBKB	123.7	98	94	Immunodeficiency 15, 615592
IKBKG	52.9	84	73	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	65.7	99	97	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IMPAD1	147.2	99	99	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INPPL1	123.8	96	93	Opsismodysplasia, 258480
KIAA0753	123.9	99	98	?Orofaciodigital syndrome XV, 617127
KIF22	161.8	100	99	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	85.8	93	88	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydroletharus syndrome 2, 614120
KMT2A	152.5	99	98	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KRAS	64.6	99	98	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	87.9	93	83	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471
LEMD3	96.5	95	88	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700

LEPRE1	137.1	100	99	Osteogenesis imperfecta, type VIII, 610915
LFNG	92.4	85	82	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	87.8	97	83	Pituitary hormone deficiency, combined, 3, 221750
LHX4	145.2	100	99	Pituitary hormone deficiency, combined, 4, 262700
LIFR	123.3	97	92	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMX1B	111.5	96	92	Nail-patella syndrome, 161200
LONP1	141.7	98	96	CODAS syndrome, 600373
LRP4	166.8	99	98	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LRP5	190.1	98	97	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRRK1	155.5	98	97	No OMIM phenotype Osteosclerotic metaphyseal dysplasia (Iida (2016) J Med Genet 53,568) ?Parkinson disease (Schulte (2013) Neurogenetics epub,epub)
LTBP2	104.7	99	97	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	113.8	98	94	Dental anomalies and short stature, 601216
LZTR1	134.3	100	99	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MAN2B1	122.5	99	96	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99	97	Mannosidosis, beta, 248510
MAP2K1	92.3	99	95	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	108	97	89	Cardiofaciocutaneous syndrome 4, 615280
MAP3K7	114.5	99	98	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137

MATN3	116.7	84	84	Epiphyseal dysplasia, multiple, 5, 607078 Spondyloepimetaphyseal dysplasia, 608728 {Osteoarthritis susceptibility 2}, 140600
MEOX1	76.8	96	91	Klippel-Feil syndrome 2, 214300
MESP2	81.8	93	87	Spondylocostal dysostosis 2, autosomal recessive, 608681
MGP	131.8	92	91	Keutel syndrome, 245150
MMP13	123.6	93	91	Metaphyseal anadysplasia 1, 602111 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	152.4	100	99	?Winchester syndrome, 277950
MMP2	164.8	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	125.6	96	91	Metaphyseal anadysplasia 2, 613073
MTAP	109.2	93	86	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYH3	110.4	99	98	Arthrogyrosis, distal, type 2A, 193700 Arthrogyrosis, distal, type 2B, 601680 Arthrogyrosis, distal, type 8, 178110
NAGLU	108.9	92	90	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NANS	106.1	100	99	Sponyloepimetaphyseal dysplasia, Genevieve type, 610442
NBAS	145.3	99	97	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEK1	103	98	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	136.8	99	98	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogyrosis, Perthes disease, and upward gaze palsy, 614262
NEU1	18.5	12	12	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NIN	144.4	99	98	Seckel syndrome 7, 614851
NKX3-2	55.7	92	73	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NOTCH2	172.7	100	99	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPC	88.1	99	96	
NPR2	165	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255

NRAS	188.2	100	100	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 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
OBSL1	140.8	99	98	3-M syndrome 2, 612921
OFD1	53	84	68	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
ORC1	106.7	99	98	Meier-Gorlin syndrome 1, 224690
ORC4	57.5	95	82	Meier-Gorlin syndrome 2, 613800
ORC6	126.6	100	100	Meier-Gorlin syndrome 3, 613803
OSTM1	80.8	90	88	Osteopetrosis, autosomal recessive 5, 259720
OTX2	155.1	100	99	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P4HB	105.3	94	94	Cole-Carpenter syndrome 1, 112240
PAM16	50.8	65	64	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	175.5	100	99	No OMIM phenotype Short stature (Dauber (2016) EMBO Mol Med epub,epub)
PAPSS2	108.4	99	98	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PCNT	117.7	99	96	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	113.6	98	94	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE4D	106	96	92	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PEX5	111.8	99	98	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	113.5	89	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100

PHEX	123.8	98	96	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	118.9	100	99	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIK3R1	129.3	99	97	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PITX1	145.1	91	86	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	147.9	99	97	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PLEKHM1	69.2	49	48	Osteopetrosis, autosomal recessive 6, 611497
PLK4	145.6	99	96	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD2	108.5	94	88	Bruck syndrome 2, 609220
PLS3	136.5	99	98	Bone mineral density QTL18, osteoporosis, 300910
POC1A	134.1	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POP1	114.2	100	99	Anauxetic dysplasia 2, 617396
POU1F1	106.2	98	94	Pituitary hormone deficiency, combined, 1, 613038
PPIB	118.7	100	100	Osteogenesis imperfecta, type IX, 259440
PRKAR1A	90.6	99	93	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PROKR2	332	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	76.6	91	84	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	53.3	91	75	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PTDSS1	127.2	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	108.6	99	98	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400

PTPN11	103.4	97	92	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAB33B	233.7	100	100	Smith-McCort dysplasia 2, 615222
RAF1	127.4	100	99	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	97.5	99	98	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	106.6	100	99	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	89.2	94	86	Adams-Oliver syndrome 3, 614814
RIPPLY2	63.8	99	83	?Spondylocostal dysostosis 6, 616566
RIT1	165.5	100	100	Noonan syndrome 8, 615355
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNPC3	41.3	84	60	No OMIM phenotype Growth hormone deficiency (Argente (2014) EMBO Mol Med epub, epub)
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROR2	166.2	99	98	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	126.2	96	93	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	86.6	99	93	{Autism, susceptibility to, X-linked 5}, 300847
RSPRY1	168.9	100	99	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	107.1	74	74	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	212.4	100	99	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SCARF2	80.9	85	73	Van den Ende-Gupta syndrome, 600920

SEC24D	136.8	99	98	Cole-Carpenter syndrome 2, 616294
SERPINF1	101.7	100	99	Osteogenesis imperfecta, type VI, 613982
SERPINH1	183.6	100	99	?Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SGSH	129.3	95	93	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	140.4	100	99	Frank-ter Haar syndrome, 249420
SHOC2	140.4	100	99	Noonan-like syndrome with loose anagen hair, 607721
SHOX	29.2	73	61	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SLC17A5	119.7	96	92	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC25A24	115.7	98	96	Fontaine progeroid syndrome ,612289
SLC26A2	233	100	100	Achondrogenesis Ib, 600972 Atelosteogenesis II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC29A3	203.8	99	99	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	105.8	98	94	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35D1	113.1	96	90	Schneckenbecken dysplasia, 269250
SLC39A13	114.9	99	98	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
SLCO2A1	110.8	100	99	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	169.1	99	97	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95)
SMAD4	121.2	99	98	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMARCAL1	134.9	100	99	Schimke immunoosseous dysplasia, 242900
SNRPB	76.1	99	97	Cerebrocostomandibular syndrome, 117650
SNX10	114.3	100	99	Osteopetrosis, autosomal recessive 8, 615085
SOS1	95.5	96	90	Noonan syndrome 4, 610733

				?Fibromatosis, gingival, 1, 135300
SOS2	97	98	92	Noonan syndrome 9, 616559
SOST	112.6	100	99	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX2	129.2	98	93	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	37.9	86	71	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	134.2	97	93	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290 Campomelic dysplasia, 114290
SP7	159.3	99	99	?Osteogenesis imperfecta, type XII, 613849
SPARC	161	100	100	Osteogenesis imperfecta, type XVII, 616507
SPINK5	145	99	96	Atopy, 147050 Netherton syndrome, 256500
SPR	166.9	98	89	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SRCAP	154.2	99	99	Floating-Harbor syndrome, 136140
STAT3	119.6	99	99	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	130.7	99	97	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
SULF1	164.5	100	99	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95) ?Hyperinsulinism (Proverbio (2013) PLoS One 8,e68740)
SUMF1	106.4	99	92	Multiple sulfatase deficiency, 272200
TAPT1	83.6	88	85	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelink type, 616897
TBCE	127.9	99	98	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBX15	131.9	100	99	Cousin syndrome, 260660
TBX4	170.9	94	92	Ischiocoxopodopatellar syndrome, 147891
TBX6	118.6	91	78	Spondylocostal dysostosis 5, 122600
TBXAS1	140.3	100	100	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158

TCIRG1	113.7	95	89	Osteopetrosis, autosomal recessive 1, 259700
TCTEX1D2	126.4	100	99	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN2	144.2	99	97	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	127.5	100	99	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TGFB1	87.1	99	95	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TMEM165	112.2	99	97	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	112	100	98	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.8	100	99	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM38B	114.3	100	99	Osteogenesis imperfecta, type XIV, 615066
TNFRSF11A	147	93	91	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	224.9	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	150.3	99	93	Osteopetrosis, autosomal recessive 2, 259710
TRAPPC2	89	87	68	Spondyloepiphyseal dysplasia tarda, 313400
TRIP11	84.2	95	87	Achondrogenesis, type IA, 200600
TRPS1	174.7	100	99	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV4	172.7	99	98	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	100.6	99	97	Nephronophthisis 12, 613820

				Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
VDR	121.1	98	95	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
WDR19	132	99	98	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	106.7	99	96	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	145.1	99	97	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	114.2	99	96	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WISP3	115.5	100	100	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNT1	189.2	100	99	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT5A	156.1	100	100	Robinow syndrome, autosomal dominant 1, 180700
XRCC4	102.9	99	97	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	132.7	90	87	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	136.5	98	94	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
ZBTB16	151.4	100	100	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZMPSTE24	113.3	100	99	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210

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.

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 : April 14th, 2017.

This list is accurate for panel version DG 2.12

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors