SHORT STATURE/SKELETAL DYSPLASIA GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCC9	175	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ACAN	136.5	92%	85%	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACP5	231.8	100%	99%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACVR1	184.8	100%	99%	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	124.3	99%	99%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	135.3	91%	88%	Weill-Marchesani-like syndrome, 613195
ADAMTSL2	124.8	96%	91%	Geleophysic dysplasia 1, 231050
AGA	175.8	100%	100%	Aspartylglucosaminuria, 208400
AGPS	69.6	98%	91%	Rhizomelic chondrodysplasia punctata, type 3, 600121
ALG12	171.3	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG3	116.7	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG9	128.6	99%	99%	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALMS1	208.7	99%	99%	Alstrom syndrome, 203800
ALPL	164.9	100%	100%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
AMER1	102.8	99%	98%	Osteopathia striata with cranial sclerosis, 300373
ANKH	124.8	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	118.5	98%	95%	KBG syndrome, 148050
ANO5	168	99%	98%	Gnathodiaphyseal dysplasia, 166260

				Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307
ARSB	137.9	98%	94%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	107.3	98%	93%	Chondrodysplasia punctata, X-linked recessive, 302950
B3GALT6	56.4	76%	71%	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	105.3	98%	92%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	122.7	97%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BMP1	168.1	100%	99%	Osteogenesis imperfecta, type XIII, 614856
BMPER	163.9	100%	99%	Diaphanospondylodysostosis, 608022
BMPR1B	195.7	100%	99%	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600
BRAF	86.4	91%	82%	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	139.7	100%	99%	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755
C21orf2	112.5	99%	98%	No OMIM phenotype Retinal dystrophy,early-onset with macular staphyloma (Khan (2015) Br J Ophtalmol 99,1725) Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236) Jeune syndrome (Wheway (2015) Nat Cell Biol 17,1074)
CA2	178.8	99%	97%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CANT1	165.9	100%	99%	Desbuquois dysplasia 1, 251450
CBL	146	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CCDC8	117.6	100%	100%	3-M syndrome 3, 614205
CDC45	183.8	99%	97%	Meier-Gorlin syndrome 7, 617063
CDC6	172.4	100%	99%	?Meier-Gorlin syndrome 5, 613805
CDKN1C	37.4	77%	63%	Beckwith-Wiedemann syndrome, 130650

				IMAGE syndrome, 614732
CDT1	106	98%	94%	Meier-Gorlin syndrome 4, 613804
CEP120	163	100%	99%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CHST3	107.5	99%	98%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CLCN5	149.8	99%	98%	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	156.7	99%	97%	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
COG1	129.4	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COL10A1	116.3	99%	97%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	112.4	96%	92%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	14.6	59%	23%	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	146	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	118.4	97%	94%	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	119.3	99%	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 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 sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL9A1	143.9	99%	97%	Stickler syndrome, type IV, 614134 /?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	77.1	99%	94%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284 {Intervertebral disc disease, susceptibility to}, 603932
COL9A3	78.3	97%	91%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	221.8	100%	99%	3MC syndrome 2, 265050
COMP	143.3	96%	93%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
CREB3L1	131.6	99%	96%	No OMIM phenotype Osteogenesis imperfecta, autosomal recessive (Symoens (2013) Orphanet J Rare Dis 8)
CRTAP	132.5	99%	97%	Osteogenesis imperfecta, type VII, 610682
CSGALNACT1	229.8	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	153.2	99%	99%	Galactosialidosis, 256540

CTSK	118.2	100%	99%	Pycnodysostosis, 265800
CUL7	166.4	99%	98%	3-M syndrome 1, 273750
CYP26B1	188	100%	99%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	147.5	100%	99%	Vitamin D-dependent rickets, type I, 264700
DDR2	172.2	100%	99%	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DHCR24	205.7	100%	100%	Desmosterolosis, 602398
DLL3	82.5	92%	83%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX3	128.9	99%	96%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DMP1	151.2	99%	99%	Hypophosphatemic rickets, AR, 241520
DONSON	119.8	92%	85%	No OMIM phenotype Microcephalic dwarfism (Reynolds (2017) Nat Genet 49,537)
DPM1	149.2	92%	87%	Congenital disorder of glycosylation, type Ie, 608799
DVL1	119	99%	96%	Robinow syndrome, autosomal dominant 2, 616331
DYM	116.7	97%	96%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC2H1	110.6	98%	91%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EBP	89.2	99%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EIF2AK3	177.3	96%	91%	Wolcott-Rallison syndrome, 226980
ENPP1	174.9	94%	88%	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	126.1	94%	91%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	137.8	96%	94%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXT1	105.5	99%	97%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	178.8	99%	99%	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
EXTL3	221.3	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425

FAM111A	292.7	100%	99%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM20C	118.7	99%	98%	Raine syndrome, 259775
FBN1	177.6	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	139.2	99%	98%	Leukocyte adhesion deficiency,type III,612840
FGD1	98.4	94%	88%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF23	122.5	99%	98%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF8	137.4	87%	80%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	165.2	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	155.6	97%	96%	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 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 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	129.1	100%	99%	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	214.4	100%	99%	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691
FKBP10	177.3	98%	95%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FLNA	160.7	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	169.5	100%	99%	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FN1	156.3	100%	99%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FUCA1	151.4	100%	99%	Fucosidosis, 230000
FZD2	200.5	99%	97%	No OMIM phenotype Omodysplasia, autosomal dominant (Saal (2015) Hum Mol Genet 24,3399)
GALNS	108.4	99%	95%	Mucopolysaccharidosis IVA, 253000
GALNT3	166.7	99%	98%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GDF3	137.7	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	179.6	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	88.9	98%	92%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GH1	229.6	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	245.4	99%	99%	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone

				Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	148.2	100%	99%	Growth hormone deficiency, isolated, type IB, 612781
GHSR	198.6	99%	98%	Growth hormone deficiency, isolated partial, 615925
GJA1	238.2	100%	100%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia 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	97.3	99%	97%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLI2	148.1	99%	97%	Culler-Jones syndrome, 615849 Holoprosencephaly-9, 610829
GLI3	168.4	99%	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
GNAS	154.4	99%	97%	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	176.5	99%	97%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	190.4	99%	97%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	169.7	96%	92%	Mucolipidosis III gamma, 252605

GNS	119.6	97%	93%	Mucopolysaccharidosis type IIID, 252940
GORAB	203.4	99%	97%	Geroderma osteodysplasticum, 231070
GPC6	160	100%	100%	Omodysplasia 1, 258315
GPR161	209.5	100%	100%	No OMIM phenotype Pituitary stalk interuption syndrome (Karaca (2015) J Clin Endocrinol Metab 100,E140)
GPX4	195.5	87%	85%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GUSB	126.8	91%	87%	Mucopolysaccharidosis VII, 253220
HDAC4	121.1	100%	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	44.3	79%	66%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	96.7	99%	96%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HGSNAT	122.6	81%	81%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HMGA2	88.3	82%	76%	Leiomyoma, uterine, somatic, 150699
HPGD	110.1	99%	98%	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HRAS	204.3	100%	99%	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	105.5	93%	87%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPG2	133.9	99%	98%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	174.4	100%	100%	Hydrolethalus syndrome, 236680
IDH1	116.2	95%	87%	{Glioma, susceptibility to, somatic}, 137800
IDH2	108	99%	98%	D-2-hydroxyglutaric aciduria 2, 613657

IDS	113.9	99%	97%	Mucopolysaccharidosis II, 309900
IDUA	120.5	92%	86%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFITM5	84.9	99%	97%	Osteogenesis imperfecta, type V, 610967
IFT122	165.7	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	124.5	99%	98%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	125.7	100%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	128	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	79.6	92%	79%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IGF1	157.6	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	153.4	100%	99%	Insulin-like growth factor I, resistance to, 270450
IGF2	110.7	100%	100%	?Growth restriction, severe, with distinctive facies, 616489
IGFALS	83.7	100%	99%	Acid-labile subunit, deficiency of, 615961
IGSF1	96.1	99%	96%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	147.1	100%	100%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	129.9	98%	95%	Immunodeficiency 15, 615592
IKBKG	57.4	83%	72%	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	75.4	99%	97%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IMPAD1	152.8	100%	99%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INPPL1	142.3	98%	95%	Opsismodysplasia, 258480
KIF22	179.3	100%	99%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	95.3	95%	89%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120

KRAS	89.6	99%	99%	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	97.8	95%	90%	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471
LEMD3	116.1	98%	93%	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700
LEPRE1	149.8	100%	99%	Osteogenesis imperfecta, type VIII, 610915
LFNG	99.3	84%	82%	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	107.4	98%	91%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	158.6	100%	99%	Pituitary hormone deficiency, combined, 4, 262700
LIFR	156.7	98%	95%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMX1B	131.5	99%	96%	Nail-patella syndrome, 161200
LONP1	164.9	98%	97%	CODAS syndrome, 600373
LRP4	184.3	99%	98%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LRP5	210	98%	98%	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	171.5	99%	98%	No OMIM phenotype

				Osteosclerotic metaphyseal dysplasia (Iida (2016) J Med Genet 53,568) ?Parkinson disease (Schulte (2013) Neurogenetics epub,epub)
LTBP2	121.8	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	128.7	98%	97%	Dental anomalies and short stature, 601216
LZTR1	166.5	100%	99%	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MAN2B1	134.6	98%	95%	Mannosidosis, alpha-, types I and II, 248500
MANBA	153.6	99%	98%	Mannosidosis, beta, 248510
MAP2K1	106.2	99%	97%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	122.2	99%	95%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K7	142.8	99%	98%	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MATN3	125.7	84%	84%	Epiphyseal dysplasia, multiple, 5, 607078 Spondyloepimetaphyseal dysplasia, 608728 {Osteoarthritis susceptibility 2}, 140600
MEOX1	110.2	98%	95%	Klippel-Feil syndrome 2, 214300
MESP2	82.3	93%	86%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MGP	161.6	95%	92%	Keutel syndrome, 245150
MMP13	145.7	92%	92%	Metaphyseal anadysplasia 1, 602111 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	166.9	100%	99%	?Winchester syndrome, 277950
MMP2	173.5	100%	99%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	134.7	99%	97%	Metaphyseal anadysplasia 2, 613073
MTAP	113.9	95%	88%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYH3	123.5	99%	98%	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
NAGLU	131.8	94%	91%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NBAS	176.4	99%	98%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800

NEK1	142.4	99%	96%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	150	99%	98%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	20.4	72%	43%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NIN	173	99%	99%	Seckel syndrome 7, 614851
NKX3-2	60.3	90%	71%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NOTCH2	180.3	100%	99%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPR2	178.1	100%	99%	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NRAS	203.3	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	152.4	99%	97%	3-M syndrome 2, 612921
OFD1	59.2	87%	75%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
ORC1	129.5	99%	98%	Meier-Gorlin syndrome 1, 224690
ORC4	81.4	97%	89%	Meier-Gorlin syndrome 2, 613800
ORC6	152.4	100%	100%	Meier-Gorlin syndrome 3, 613803
OSTM1	96.5	92%	89%	Osteopetrosis, autosomal recessive 5, 259720
OTX2	140.5	100%	99%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P4HB	123.6	94%	94%	Cole-Carpenter syndrome 1, 112240
PAM16	57.4	65%	64%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320

PAPPA2	185	99%	99%	No OMIM phenotype Short stature (Dauber (2016) EMBO Mol Med epub,epub)
PAPSS2	113.6	99%	99%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PCNT	128.9	99%	96%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	131.4	99%	96%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE4D	132.3	98%	96%	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PEX5	132.1	99%	98%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX7	152.7	90%	87%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	138.1	98%	97%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	138	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIK3R1	158.1	99%	98%	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PITX2	146.1	99%	98%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PLEKHM1	68	90%	86%	Osteopetrosis, autosomal recessive 6, 611497
PLK4	172.8	99%	97%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD2	143.1	97%	92%	Bruck syndrome 2, 609220
PLS3	156.3	99%	99%	Bone mineral density QTL18, osteoporosis, 300910
POC1A	144.7	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POP1	131.6	99%	99%	Anauxetic dysplasia 2, 617396
POU1F1	141.9	99%	97%	Pituitary hormone deficiency, combined, 1, 613038
PPIB	148.1	100%	100%	Osteogenesis imperfecta, type IX, 259440
PRKAR1A	104.2	97%	92%	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	387.8	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	92.2	94%	86%	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	53.4	91%	77%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PTDSS1	162.9	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	118.4	99%	98%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPN11	105.7	98%	93%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAB33B	282.1	100%	100%	Smith-McCort dysplasia 2, 615222
RAF1	144.9	100%	99%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	109.7	99%	98%	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	109.4	99%	97%	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	94.2	95%	88%	Adams-Oliver syndrome 3, 614814
RIT1	190.3	100%	100%	Noonan syndrome 8, 615355
RMRP	999999			Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNPC3	54.1	92%	74%	No OMIM phenotype Growth hormone deficiency (Argente (2014) EMBO Mol Med epub, epub)
RNU4ATAC	999999			Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROR2	190.4	99%	99%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	160.8	96%	95%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPL10	94.2	98%	91%	{Autism, susceptibility to, X-linked 5}, 300847
RSPRY1	186.1	100%	100%	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type, 616723

RUNX2	111.8	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	231.2	100%	99%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SCARF2	88.8	94%	84%	Van den Ende-Gupta syndrome, 600920
SEC24D	159.3	99%	98%	Cole-Carpenter syndrome 2, 616294
SERPINF1	117.3	100%	99%	Osteogenesis imperfecta, type VI, 613982
SERPINH1	188	100%	99%	?Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SGSH	140.9	96%	94%	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900
SH3PXD2B	158.6	100%	99%	Frank-ter Haar syndrome, 249420
SHOC2	150.5	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SHOX	28.6	78%	59%	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SLC17A5	142.3	99%	95%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC26A2	289.4	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	236.3	99%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	120.6	99%	96%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35D1	144.4	97%	93%	Schneckenbecken dysplasia, 269250
SLC39A13	134.6	99%	99%	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
SLCO2A1	111.8	99%	98%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	179.5	99%	99%	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95)
SMAD4	136.5	99%	99%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350

				Polyposis, juvenile intestinal, 174900
SMARCAL1	153.8	100%	99%	Schimke immunoosseous dysplasia, 242900
SNRPB	91	99%	97%	Cerebrocostomandibular syndrome, 117650
SNX10	142	100%	99%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	120.1	98%	95%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	115.8	99%	97%	Noonan syndrome 9, 616559
SOST	133.2	100%	99%	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX2	127.1	99%	98%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	47	94%	81%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	138.7	97%	93%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290 Campomelic dysplasia, 114290
SP7	173.4	99%	99%	?Osteogenesis imperfecta, type XII, 613849
SPARC	172.7	100%	100%	Osteogenesis imperfecta, type XVII, 616507
SPINK5	182.2	99%	98%	Atopy, 147050 Netherton syndrome, 256500
SPR	194	99%	90%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SRCAP	165.3	99%	98%	Floating-Harbor syndrome, 136140
STAT3	132.1	100%	99%	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	140.5	99%	96%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
SULF1	178	100%	99%	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95) ?Hyperinsulinism (Proverbio (2013) PLoS One 8,e68740)
SUMF1	137.1	99%	94%	Multiple sulfatase deficiency, 272200
TAPT1	113.6	89%	86%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type,616897
TBCE	162.8	99%	99%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460

TBX15	127.2	100%	99%	Cousin syndrome, 260660
TBX4	198	96%	94%	Ischiocoxopodopatellar syndrome, 147891
TBX6	132.1	96%	86%	Spondylocostal dysostosis 5, 122600
TBXAS1	170.1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
TCIRG1	127.8	96%	89%	Osteopetrosis, autosomal recessive 1, 259700
TCTEX1D2	139.6	100%	100%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN2	166.2	99%	97%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	133.6	100%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TGFB1	84.7	99%	98%	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700
TMEM165	136.2	99%	98%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	147.1	99%	99%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	105.5	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM38B	124.3	100%	100%	Osteogenesis imperfecta, type XIV, 615066
TNFRSF11A	152.6	94%	91%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	258.6	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	177.2	99%	96%	Osteopetrosis, autosomal recessive 2, 259710
TRAPPC2	113.4	91%	77%	Spondyloepiphyseal dysplasia tarda, 313400
TRIP11	114.2	97%	92%	Achondrogenesis, type IA, 200600
TRPS1	196.7	100%	100%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV4	188.6	99%	99%	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	132.4	99%	98%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
VDR	124.6	99%	96%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
WDR19	170	100%	99%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	122	99%	96%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	186.3	99%	98%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	131.2	99%	97%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WISP3	132	100%	99%	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNT1	225.8	100%	99%	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT5A	181.5	100%	99%	Robinow syndrome, autosomal dominant 1, 180700
XRCC4	134.1	99%	98%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	144.5	92%	88%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	161.9	98%	96%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
ZBTB16	165.9	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZMPSTE24	151.9	99%	99%	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 : April 14th 2017
This list is accurate for panel version DG 2.9

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