SHORT STATURE AND SKELETAL DYSPLASIA GENE PANEL DG 2.17 (521 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ABCC9	140.4	100.0%	99.9%	Hypertrichotic osteochondrodysplasia, 239850 Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ACAN	132.5	96.0%	90.1%	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACP5	189.0	100.0%	99.9%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVR1	139.8	100.0%	100.0%	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	135.4	100.0%	99.9%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	118.3	98.7%	93.9%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	126.3	99.1%	96.8%	Geleophysic dysplasia 1, 231050
AGA	144.3	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGPS	74.8	100.0%	97.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AIFM1	92.9	99.7%	96.5%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
ALG12	169.5	100.0%	100.0%	Congenital disorder of glycosylation, type Ig, 607143
ALG3	117.9	100.0%	100.0%	Congenital disorder of glycosylation, type Id, 601110
ALG9	114.8	100.0%	99.8%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	178.0	100.0%	99.8%	Alstrom syndrome, 203800
ALPL	168.4	99.9%	99.5%	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300

				Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALX1	139.5	100.0%	99.2%	?Frontonasal dysplasia 3, 613456
ALX3	148.9	91.7%	80.3%	Frontonasal dysplasia 1, 136760
ALX4	175.4	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMER1	106.2	99.8%	99.1%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	102.3	100.0%	99.6%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKH	116.6	100.0%	100.0%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	131.8	99.6%	97.6%	KBG syndrome, 148050
ANO5	126.7	99.7%	97.1%	Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANTXR2	117.5	99.8%	97.7%	Hyaline fibromatosis syndrome, 228600
APC2	142.2	100.0%	99.6%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
ARHGAP31	151.5	99.9%	99.1%	Adams-Oliver syndrome 1, 100300
ARID1B	150.6	99.5%	99.3%	Coffin-Siris syndrome 1, 135900
ARSB	111.3	100.0%	99.4%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	83.5	98.5%	91.1%	Chondrodysplasia punctata, X-linked recessive, 302950
ATP6V0A2	120.5	100.0%	99.6%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATR	142.1	99.9%	98.9%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
B3GALT6	96.5	87.5%	80.2%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	134.2	99.9%	97.0%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	138.9	100.0%	99.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BGN	147.7	100.0%	100.0%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	41.5	97.8%	83.4%	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BMP1	167.3	100.0%	100.0%	Osteogenesis imperfecta, type XIII, 614856
BMP2	180.6	100.0%	100.0%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877

BMPER	132.4	100.0%	99.6%	Diaphanospondylodysostosis, 608022
BMPR1B	141.6	100.0%	100.0%	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
BRAF	71.0	91.7%	79.4%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRF1	120.2	99.9%	98.8%	Cerebellofaciodental syndrome, 616202
BTK	98.6	100.0%	99.1%	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 Agammaglobulinemia, X-linked 1, 300755
BTRC	132.6	98.9%	97.4%	No OMIM Disease ID
C16orf62	143.8	100.0%	99.9%	No OMIM Disease ID
C21orf2	146.9	100.0%	99.4%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
C5orf42	122.3	99.7%	97.4%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	141.8	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CANT1	158.4	100.0%	100.0%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CASR	167.3	100.0%	99.8%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CBL	131.1	97.4%	97.1%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CC2D2A	112.6	99.0%	97.0%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCDC8	211.7	100.0%	100.0%	3-M syndrome 3, 614205
CDC42	89.8	98.1%	90.1%	Takenouchi-Kosaki syndrome, 616737
CDC45	148.0	99.6%	98.4%	Meier-Gorlin syndrome 7, 617063
CDC6	142.2	100.0%	99.7%	?Meier-Gorlin syndrome 5, 613805
CDC73	111.0	100.0%	98.9%	Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001

				Parathyroid carcinoma, 608266 Hyperparathyroidism, familial primary, 145000
CDKN1C	116.5	93.6%	84.7%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDT1	150.4	100.0%	100.0%	Meier-Gorlin syndrome 4, 613804
CENPE	72.2	98.5%	91.7%	?Microcephaly 13, primary, autosomal recessive, 616051
CEP120	131.3	100.0%	99.6%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP152	145.3	99.6%	97.8%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	77.6	96.9%	88.7%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CHST14	180.8	100.0%	99.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	146.9	100.0%	100.0%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHSY1	134.8	99.8%	99.0%	Temtamy preaxial brachydactyly syndrome, 605282
CKAP2L	154.2	99.9%	98.9%	Filippi syndrome, 272440
CLCN5	105.8	99.7%	96.8%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
CLCN7	162.0	99.9%	98.9%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
COG1	117.9	100.0%	99.9%	Congenital disorder of glycosylation, type IIg, 611209
COG4	99.0	100.0%	99.7%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COL10A1	116.2	100.0%	100.0%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	94.6	98.0%	93.6%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
COL11A2	122.3	100.0%	99.5%	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524

				Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL1A1	154.6	99.9%	99.1%	Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL1A2	96.3	98.6%	94.6%	Ehlers-Danlos syndrome, cardiac valvular type, 225320 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL27A1	154.1	99.9%	99.3%	Steel syndrome, 615155
COL2A1	121.1	100.0%	99.8%	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 SED congenita, 183900 Legg-Calve-Perthes disease, 150600 SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL9A1	132.9	100.0%	99.5%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	104.9	100.0%	99.6%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	120.1	99.9%	98.3%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
COLEC11	197.8	100.0%	100.0%	3MC syndrome 2, 265050
COMP	147.0	97.4%	93.9%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
CREB3L1	145.4	100.0%	99.9%	Osteogenesis imperfecta, type XVI, 616229

CREBBP	120.9	99.6%	97.3%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRIPT	40.5	99.0%	89.7%	Short stature with microcephaly and distinctive facies, 615789
CRTAP	129.9	100.0%	99.3%	Osteogenesis imperfecta, type VII, 610682
CSF1R	121.9	100.0%	99.4%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSGALNACT1	161.0	100.0%	99.8%	No OMIM Disease ID
CTSA	146.1	100.0%	100.0%	Galactosialidosis, 256540
CTSK	90.0	100.0%	100.0%	Pycnodysostosis, 265800
CUL7	139.6	100.0%	100.0%	3-M syndrome 1, 273750
CYP26B1	188.5	100.0%	100.0%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	164.1	100.0%	99.8%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	133.4	99.9%	97.5%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
DDR2	119.0	100.0%	99.7%	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	108.7	100.0%	99.9%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX58	111.6	99.9%	98.4%	Singleton-Merten syndrome 2, 616298
DHCR24	170.7	100.0%	99.9%	Desmosterolosis, 602398
DHODH	107.2	100.0%	100.0%	Miller syndrome, 263750
DLL3	122.2	97.9%	93.8%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	194.6	100.0%	100.0%	Adams-Oliver syndrome 6, 616589
DLX3	165.9	100.0%	99.8%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX5	159.7	100.0%	99.8%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	140.6	100.0%	100.0%	No OMIM Disease ID
DMP1	135.8	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
DNA2	121.9	99.9%	97.8%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJC21	130.6	100.0%	99.2%	Bone marrow failure syndrome 3, 617052
DNMT3A	132.4	99.9%	98.7%	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DOCK6	132.3	99.6%	98.9%	Adams-Oliver syndrome 2, 614219
DONSON	92.4	99.6%	94.7%	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPCD	128.0	100.0%	100.0%	No OMIM Disease ID
DPM1	134.2	95.5%	87.7%	Congenital disorder of glycosylation, type Ie, 608799
DSE	95.2	99.8%	98.0%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539

DVL1	159.7	99.3%	97.2%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	209.9	100.0%	100.0%	Robinow syndrome, autosomal dominant 3, 616894
DYM	102.4	97.4%	95.7%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC2H1	98.0	98.9%	94.3%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	94.8	99.6%	97.1%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EBP	68.9	99.8%	96.3%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EDN1	165.0	100.0%	99.9%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798
EDNRA	153.6	100.0%	99.9%	Mandibulofacial dysostosis with alopecia, 616367
EFL1	152.7	99.4%	98.0%	Shwachman-Diamond syndrome 2, 617941
EFNB1	122.4	100.0%	100.0%	Craniofrontonasal dysplasia, 304110
EFTUD2	107.1	100.0%	99.5%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2AK3	134.0	99.5%	96.7%	Wolcott-Rallison syndrome, 226980
EIF4A3	89.6	100.0%	98.9%	Robin sequence with cleft mandible and limb anomalies, 268305
ENPP1	128.6	97.9%	92.4%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
EOGT	103.3	79.5%	78.1%	Adams-Oliver syndrome 4, 615297
EP300	173.3	99.7%	98.7%	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
ERF	166.3	100.0%	99.6%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ESCO2	112.4	99.5%	96.2%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
EVC	113.0	96.8%	92.1%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	115.9	99.6%	97.1%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EXOC6B	107.7	98.5%	97.1%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXT1	91.1	99.9%	98.4%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	120.9	99.9%	99.0%	Exostoses, multiple, type 2, 133701 Seizures, scoliosis, and macrocephaly syndrome, 616682
EXTL3	200.7	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425

EZH2	130.3	99.5%	98.0%	Weaver syndrome, 277590
FAM111A	241.7	100.0%	99.6%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM20B	132.5	100.0%	99.8%	No OMIM Disease ID
FAM20C	165.1	100.0%	100.0%	Raine syndrome, 259775
FAM46A	177.0	100.0%	100.0%	Osteogenesis imperfecta, type XVIII, 617952
FAM58A	57.8	83.7%	78.2%	STAR syndrome, 300707
FAR1	72.4	97.6%	91.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBLN1	146.0	99.8%	98.7%	No OMIM Disease ID
FBN1	138.3	100.0%	99.7%	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FBN2	144.5	100.0%	99.9%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXW4	108.1	100.0%	99.8%	No OMIM Disease ID
FERMT3	161.2	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FGD1	93.2	98.7%	94.4%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	118.7	100.0%	99.7%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	130.1	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF8	141.1	98.8%	90.5%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	156.9	100.0%	100.0%	Multiple synostoses syndrome 3, 612961
FGFR1	131.6	100.0%	99.7%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	118.0	97.7%	97.1%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150

				Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	157.1	100.0%	99.9%	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FIG4	155.0	100.0%	99.7%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FKBP10	170.1	99.8%	98.3%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	77.6	100.0%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLNA	156.4	100.0%	99.9%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350

				Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FLNB	131.8	99.8%	99.1%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FMN1	130.5	99.0%	97.6%	No OMIM Disease ID
FN1	109.6	100.0%	99.3%	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FUCA1	135.9	100.0%	100.0%	Fucosidosis, 230000
FUZ	143.2	100.0%	100.0%	No OMIM Disease ID
FZD2	200.1	99.9%	98.5%	Omodysplasia 2, 164745
GALNS	118.1	100.0%	99.4%	Mucopolysaccharidosis IVA, 253000
GALNT3	126.0	99.9%	98.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GCM2	142.3	100.0%	100.0%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GDF3	132.5	100.0%	100.0%	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702
GDF5	190.2	100.0%	100.0%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A1, C, 615072 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A2, 112600 Du Pan syndrome, 228900 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017
GDF6	180.6	100.0%	100.0%	Leber congenital amaurosis 17, 615360 Klippel-Feil syndrome 1, autosomal dominant, 118100 Multiple synostoses syndrome 4, 617898 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GH1	167.8	100.0%	100.0%	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type IA, 262400

				Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100
GHR	152.0	99.8%	99.8%	Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 Growth hormone insensitivity, partial, 604271
GHRHR	115.5	96.3%	95.4%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	194.4	98.7%	95.5%	Growth hormone deficiency, isolated partial, 615925
GJA1	162.4	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GLB1	87.4	99.5%	95.2%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLI2	177.4	100.0%	100.0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	151.8	100.0%	99.5%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GMNN	123.0	99.7%	94.8%	Meier-Gorlin syndrome 6, 616835
GNAI3	88.5	98.9%	93.5%	Auriculocondylar syndrome 1, 602483
GNAS	241.4	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580
GNPAT	128.8	99.7%	96.6%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	149.5	100.0%	99.4%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600

GNPTG	199.0	99.9%	99.4%	Mucolipidosis III gamma, 252605
GNS	93.6	99.9%	97.2%	Mucopolysaccharidosis type IIID, 252940
GORAB	168.3	100.0%	99.1%	Geroderma osteodysplasticum, 231070
GPC3	76.8	98.9%	93.5%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	132.5	100.0%	100.0%	Omodysplasia 1, 258315
GPR161	181.5	100.0%	100.0%	No OMIM Disease ID
GPX4	185.8	95.2%	91.9%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GSC	149.2	100.0%	99.6%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GUSB	106.6	92.6%	91.1%	Mucopolysaccharidosis VII, 253220
GZF1	205.4	100.0%	99.6%	Joint laxity, short stature, and myopia, 617662
HAAO	113.2	100.0%	100.0%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HDAC4	131.1	100.0%	100.0%	No OMIM Disease ID
HDAC8	110.1	100.0%	99.6%	Cornelia de Lange syndrome 5, 300882
HES7	61.9	93.1%	78.9%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	65.7	99.9%	97.5%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	99.9	88.2%	86.3%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HMGA2	85.9	82.3%	75.8%	No OMIM Disease ID
HOXA11	96.7	99.8%	97.7%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	90.2	95.8%	84.5%	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXD13	222.6	100.0%	100.0%	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 ?Brachydactyly-syndactyly syndrome, 610713 Syndactyly, type V, 186300 Synpolydactyly 1, 186000
HPGD	87.8	100.0%	99.7%	Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HRAS	196.0	100.0%	100.0%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200

				Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HSPA9	83.8	88.2%	84.2%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPG2	132.7	99.5%	99.2%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	160.4	100.0%	100.0%	Hydrolethalus syndrome, 236680
IARS2	145.7	100.0%	100.0%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
ICK	111.0	100.0%	98.8%	Endocrine-cerebroosteodysplasia, 612651
ID4	203.9	100.0%	99.0%	No OMIM Disease ID
IDH1	80.0	90.8%	78.1%	No OMIM Disease ID
IDH2	107.4	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDS	105.1	99.8%	97.2%	Mucopolysaccharidosis II, 309900
IDUA	169.2	99.3%	96.4%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IFIH1	110.9	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	104.5	100.0%	99.6%	Osteogenesis imperfecta, type V, 610967
IFT122	126.6	100.0%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT140	127.6	100.0%	99.6%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	98.4	100.0%	99.5%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	119.5	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	120.0	100.0%	99.7%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	61.7	95.6%	81.0%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	90.5	93.0%	88.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100.6	100.0%	100.0%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	123.1	100.0%	99.7%	Insulin-like growth factor I, resistance to, 270450
IGF2	131.1	100.0%	100.0%	?Growth restriction, severe, with distinctive facies, 616489
IGFALS	126.7	100.0%	100.0%	Acid-labile subunit, deficiency of, 615961
IGSF1	73.1	99.5%	95.2%	Hypothyroidism, central, and testicular enlargement, 300888

IHH	198.7	100.0%	100.0%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	116.2	99.4%	97.1%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	64.7	90.1%	80.2%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IL1RN	145.3	100.0%	99.9%	Interleukin 1 receptor antagonist deficiency, 612852
IL2RG	60.2	99.7%	94.3%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL6ST	83.7	94.9%	88.0%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IMPAD1	181.6	100.0%	100.0%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INPPL1	139.7	99.9%	98.9%	Opsismodysplasia, 258480
INTU	112.9	99.8%	98.0%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
KAT6B	162.3	99.8%	99.2%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNJ2	164.2	100.0%	100.0%	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KIAA0586	115.1	97.3%	92.6%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	117.1	99.9%	98.9%	?Orofaciodigital syndrome XV, 617127
KIF22	188.7	100.0%	99.9%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	120.4	99.3%	96.6%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KL	185.9	99.8%	98.9%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KMT2A	138.5	100.0%	99.9%	Wiedemann-Steiner syndrome, 605130
KRAS	64.0	99.8%	96.8%	Leukemia, acute myeloid, 601626 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278

				Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
LBR	104.4	97.4%	90.4%	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LBX1	227.8	100.0%	100.0%	No OMIM Disease ID
LEMD3	127.5	99.8%	97.8%	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
LFNG	132.2	95.1%	89.4%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	135.1	96.6%	96.6%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	141.9	100.0%	100.0%	Pituitary hormone deficiency, combined, 4, 262700
LIFR	108.8	99.6%	96.7%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMNA	118.2	98.3%	93.2%	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMX1B	163.9	100.0%	99.3%	Nail-patella syndrome, 161200
LONP1	164.8	100.0%	100.0%	CODAS syndrome, 600373
LPIN2	101.1	100.0%	99.7%	Majeed syndrome, 609628
LRP4	136.5	99.8%	99.1%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	183.1	99.9%	99.4%	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750

				Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634
LRRK1	157.6	99.5%	98.1%	No OMIM Disease ID
LTBP2	124.2	100.0%	99.7%	Glaucoma 3, primary congenital, D, 613086 ?Weill-Marchesani syndrome 3, recessive, 614819 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	166.1	100.0%	100.0%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	157.2	100.0%	99.9%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAFB	140.5	100.0%	100.0%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAN2B1	139.1	99.9%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MANBA	117.1	99.7%	98.1%	Mannosidosis, beta, 248510
MAP2K1	96.7	99.6%	97.1%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	139.8	99.3%	95.6%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K20	110.7	99.9%	98.8%	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAP3K7	117.5	100.0%	99.7%	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MATN3	103.0	87.1%	84.5%	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBTPS2	109.0	99.9%	98.4%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MECOM	133.4	100.0%	99.8%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MEGF8	158.7	100.0%	99.8%	Carpenter syndrome 2, 614976
MEOX1	114.4	100.0%	98.8%	Klippel-Feil syndrome 2, 214300
MESD	122.0	100.0%	100.0%	Osteogenesis imperfecta, type XX, 618644
MESP2	148.8	97.4%	96.2%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	153.7	100.0%	99.6%	Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MGP	134.2	98.6%	93.2%	Keutel syndrome, 245150

MIR140	NC	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MKS1	98.8	99.9%	98.5%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MMP13	113.0	92.9%	92.3%	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	158.2	100.0%	99.9%	?Winchester syndrome, 277950
MMP2	162.9	100.0%	100.0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	161.0	100.0%	99.1%	Metaphyseal anadysplasia 2, 613073
MNX1	65.4	81.8%	74.0%	Currarino syndrome, 176450
MSX2	111.9	100.0%	100.0%	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	100.1	99.2%	94.8%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYCN	200.6	100.0%	100.0%	Feingold syndrome 1, 164280
MYH3	100.2	100.0%	99.1%	Contractures, pterygia, and variable skeletal fusions syndrome 1B, 618469 Contractures, pterygia, and variable skeletal fusions syndrome 1A, 178110 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYO18B	132.4	100.0%	99.4%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
NAGLU	130.5	98.5%	95.6%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NANS	105.0	100.0%	99.3%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	138.4	99.9%	99.2%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEK1	111.2	99.9%	98.0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	123.4	99.9%	98.9%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	150.1	99.5%	96.5%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NF1	105.8	92.5%	89.3%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210

NFIX	195.4	100.0%	99.8%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NIN	130.6	99.9%	99.2%	?Seckel syndrome 7, 614851
NIPBL	123.0	98.9%	96.7%	Cornelia de Lange syndrome 1, 122470
NKX3-2	161.2	100.0%	99.9%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP3	146.0	100.0%	99.9%	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NOG	270.7	100.0%	100.0%	Tarsal-carpal coalition syndrome, 186570 Symphalangism, proximal, 1A, 185800 Stapes ankylosis with broad thumbs and toes, 184460 Multiple synostoses syndrome 1, 186500 Brachydactyly, type B2, 611377
NOTCH1	158.4	99.8%	99.2%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	130.5	100.0%	99.8%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPPC	150.4	100.0%	100.0%	No OMIM Disease ID
NPR2	154.2	100.0%	99.5%	Short stature with nonspecific skeletal abnormalities, 616255 Epiphyseal chondrodysplasia, Miura type, 615923 Acromesomelic dysplasia, Maroteaux type, 602875
NPR3	202.6	100.0%	100.0%	No OMIM disease ID
NRAS	145.3	100.0%	100.0%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NSD1	152.6	100.0%	99.8%	Sotos syndrome 1, 117550
NSDHL	133.7	99.9%	98.2%	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	82.1	100.0%	98.5%	Seckel syndrome 10, 617253
NXN	140.2	100.0%	100.0%	Robinow syndrome, autosomal recessive 2, 618529
OBSL1	166.0	100.0%	99.9%	3-M syndrome 2, 612921

OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
ORC1	93.8	99.9%	98.9%	Meier-Gorlin syndrome 1, 224690
ORC4	68.8	97.5%	90.4%	Meier-Gorlin syndrome 2, 613800
ORC6	130.5	100.0%	99.9%	Meier-Gorlin syndrome 3, 613803
OSTM1	113.8	97.8%	92.1%	Osteopetrosis, autosomal recessive 5, 259720
OTX2	135.4	100.0%	99.6%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P3H1	140.0	100.0%	100.0%	Osteogenesis imperfecta, type VIII, 610915
P4HB	117.0	94.6%	93.7%	Cole-Carpenter syndrome 1, 112240
PAM16	69.6	65.7%	65.3%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	148.0	100.0%	99.9%	No OMIM Disease ID
PAPSS2	107.4	99.8%	97.8%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PAX3	116.1	100.0%	99.9%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCNT	124.1	99.8%	98.2%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	97.1	99.1%	95.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE3A	127.5	100.0%	99.2%	Hypertension and brachydactyly syndrome, 112410
PDE4D	104.9	95.8%	95.0%	Acrodysostosis 2, with or without hormone resistance, 614613
PEX5	115.8	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	117.6	99.1%	93.9%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	107.9	99.7%	98.2%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGV	129.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300

PIK3R1	125.7	99.9%	98.8%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PISD	172.5	100.0%	99.9%	No OMIM Disease ID
PITX1	202.5	99.8%	97.6%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	186.2	100.0%	99.6%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PKDCC	96.0	97.4%	90.8%	No OMIM Disease ID
PLCB3	159.3	100.0%	99.3%	No OMIM Disease ID
PLCB4	102.7	99.8%	97.9%	Auriculocondylar syndrome 2, 614669
PLEKHM1	139.5	100.0%	100.0%	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLK4	145.8	99.9%	98.1%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	141.5	99.9%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	115.6	99.4%	96.2%	Bruck syndrome 2, 609220
PLS3	115.0	97.0%	95.5%	Bone mineral density QTL18, osteoporosis, 300910
POC1A	120.3	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POLE	136.9	99.9%	99.6%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLL	138.1	99.9%	97.7%	No OMIM Disease ID
POLR1A	108.5	99.9%	98.8%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	103.3	99.3%	95.4%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	186.7	91.6%	91.6%	Treacher Collins syndrome 2, 613717
POLR3A	119.8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	132.0	99.9%	98.3%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POP1	123.0	100.0%	99.2%	Anauxetic dysplasia 2, 617396
POR	195.6	99.4%	97.5%	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
POU1F1	109.5	99.9%	98.2%	Pituitary hormone deficiency, combined, 1, 613038
PPIB	118.3	100.0%	100.0%	Osteogenesis imperfecta, type IX, 259440
PPP1CB	110.7	100.0%	99.6%	Noonan syndrome-like disorder with loose anagen hair 2, 617506

PRKAR1A	80.5	98.1%	92.8%	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
PROKR2	245.3	100.0%	100.0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	107.5	93.9%	86.2%	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	46.2	91.6%	74.2%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PTDSS1	116.2	100.0%	100.0%	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	121.2	100.0%	99.5%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTHLH	138.1	99.1%	91.2%	Brachydactyly, type E2, 613382
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PYCR1	105.0	100.0%	99.0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	102.4	100.0%	99.8%	Carpenter syndrome, 201000
RAB33B	199.5	100.0%	100.0%	Smith-McCort dysplasia 2, 615222
RAC3	132.6	98.1%	95.4%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	80.8	98.0%	93.5%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAF1	111.1	100.0%	99.9%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RASGRP2	111.5	100.0%	99.8%	?Bleeding disorder, platelet-type, 18, 615888
RBBP8	117.9	100.0%	99.3%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBM8A	89.0	100.0%	98.2%	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	72.1	95.8%	87.2%	Adams-Oliver syndrome 3, 614814
RECQL4	181.4	100.0%	100.0%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400

RIPPLY2	83.9	99.9%	97.4%	?Spondylocostal dysostosis 6, 616566
RIT1	142.5	100.0%	100.0%	Noonan syndrome 8, 615355
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNPC3	40.8	86.2%	65.9%	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROR2	176.7	100.0%	99.9%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	124.2	96.8%	95.8%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPL10	68.6	98.5%	89.7%	Mental retardation, X-linked, syndromic, 35, 300998
RPL13	56.4	100.0%	97.6%	No OMIM Disease ID
RRAS	138.5	100.0%	99.4%	No OMIM Disease ID
RSPO2	134.3	96.8%	89.6%	Tetraamelia syndrome 2, 618021 ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022
RSPRY1	143.3	100.0%	100.0%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	109.5	73.6%	72.3%	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600
SALL1	127.8	99.9%	99.3%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	147.5	100.0%	98.7%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SBDS	167.5	100.0%	100.0%	Shwachman-Diamond syndrome, 260400
SCARF2	120.2	99.6%	97.7%	Van den Ende-Gupta syndrome, 600920
SEC24D	125.7	100.0%	99.6%	Cole-Carpenter syndrome 2, 616294
SERPINF1	109.4	100.0%	99.8%	Osteogenesis imperfecta, type VI, 613982
SERPINH1	216.7	100.0%	99.9%	Osteogenesis imperfecta, type X, 613848
SETD2	139.1	100.0%	99.7%	Luscan-Lumish syndrome, 616831
SF3B4	82.6	99.9%	98.5%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	136.0	100.0%	99.1%	Pyle disease, 265900
SGMS2	153.7	100.0%	100.0%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGSH	152.5	98.1%	94.9%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900

SH3BP2	154.6	92.4%	91.4%	Cherubism, 118400
SH3PXD2B	175.9	100.0%	99.8%	Frank-ter Haar syndrome, 249420
SHH	165.7	100.0%	100.0%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	136.8	100.0%	99.4%	Noonan syndrome-like with loose anagen hair, 607721
SHOX	40.6	85.4%	69.6%	Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300
SKI	149.5	100.0%	99.7%	Shprintzen-Goldberg syndrome, 182212
SLC10A7	110.2	99.9%	99.1%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC17A5	136.7	98.7%	95.1%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC25A24	133.1	99.7%	99.5%	Fontaine progeroid syndrome, 612289
SLC26A2	203.7	100.0%	99.9%	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900
SLC29A3	190.1	100.0%	99.7%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	165.6	100.0%	99.5%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35D1	126.1	99.6%	95.7%	Schneckenbecken dysplasia, 269250
SLC39A13	158.7	100.0%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLCO2A1	104.4	100.0%	98.7%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	167.2	99.8%	98.4%	No OMIM Disease ID
SMAD2	126.9	100.0%	99.5%	No OMIM Disease ID
SMAD3	138.0	100.0%	100.0%	Loeys-Dietz syndrome 3, 613795
SMAD4	109.9	100.0%	99.9%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA4	163.9	100.0%	99.6%	Coffin-Siris syndrome 4, 614609
SMARCAL1	119.6	100.0%	99.8%	Schimke immunoosseous dysplasia, 242900
SMARCB1	192.9	100.0%	100.0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608

SMARCE1	66.9	94.6%	85.4%	Coffin-Siris syndrome 5, 616938
SMC1A	93.2	99.9%	98.3%	Cornelia de Lange syndrome 2, 300590
SMC3	82.8	96.4%	89.7%	Cornelia de Lange syndrome 3, 610759
SNRPB	83.6	99.9%	98.8%	Cerebrocostomandibular syndrome, 117650
SNX10	124.3	96.2%	95.5%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	100.6	99.7%	96.7%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	98.0	99.7%	97.7%	Noonan syndrome 9, 616559
SOST	207.3	100.0%	99.5%	Sclerosteosis 1, 269500 Van Buchem disease, 239100 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX2	261.8	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	87.5	99.0%	95.2%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX9	181.5	100.0%	100.0%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP7	164.6	100.0%	99.5%	Osteogenesis imperfecta, type XII, 613849
SPARC	144.6	100.0%	100.0%	Osteogenesis imperfecta, type XVII, 616507
SPECC1L	133.2	100.0%	99.7%	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPINK5	128.1	100.0%	99.2%	Netherton syndrome, 256500
SPR	159.7	100.0%	100.0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	143.0	99.8%	98.1%	Legius syndrome, 611431
SRCAP	166.7	100.0%	99.8%	Floating-Harbor syndrome, 136140
SRP54	108.8	99.2%	94.7%	No OMIM Disease ID
STAT3	106.9	100.0%	99.4%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	119.7	99.9%	98.8%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
SULF1	138.8	100.0%	99.3%	No OMIM Disease ID
SUMF1	91.7	99.9%	97.6%	Multiple sulfatase deficiency, 272200
TAB2	171.5	100.0%	99.7%	Congenital heart defects, nonsyndromic, 2, 614980
TAPT1	87.2	99.3%	93.9%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897

TBCE	117.0	99.3%	95.6%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBX15	114.2	100.0%	100.0%	Cousin syndrome, 260660
TBX3	109.8	99.9%	98.7%	Ulnar-mammary syndrome, 181450
TBX4	187.7	99.8%	98.2%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891
TBX5	145.6	100.0%	100.0%	Holt-Oram syndrome, 142900
TBX6	135.0	99.8%	97.5%	Spondylocostal dysostosis 5, 122600
TBXAS1	135.5	100.0%	100.0%	Ghosal hematodiaphyseal syndrome, 231095
TCF12	137.8	100.0%	99.9%	Craniosynostosis 3, 615314
TCIRG1	149.6	99.6%	98.0%	Osteopetrosis, autosomal recessive 1, 259700
TCOF1	119.1	99.9%	99.3%	Treacher Collins syndrome 1, 154500
TCTEX1D2	126.1	99.9%	99.0%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN2	127.0	100.0%	99.0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	121.0	100.0%	100.0%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TGDS	84.3	99.3%	95.0%	Catel-Manzke syndrome, 616145
TGFB1	114.1	100.0%	99.5%	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
TGFB2	179.0	100.0%	99.8%	Loeys-Dietz syndrome 4, 614816
TGFB3	149.0	100.0%	100.0%	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBR1	156.6	97.3%	94.3%	Loeys-Dietz syndrome 1, 609192
TGFBR2	169.1	100.0%	100.0%	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
THPO	102.6	100.0%	99.9%	Thrombocythemia 1, 187950
TMEM165	159.2	99.9%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	92.0	99.9%	96.9%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	112.1	100.0%	99.7%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM38B	110.2	100.0%	99.1%	Osteogenesis imperfecta, type XIV, 615066
TMEM67	80.6	99.3%	93.5%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550

				COACH syndrome, 216360 Joubert syndrome 6, 610688
TNFRSF11A	139.5	96.4%	95.6%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	175.0	100.0%	100.0%	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	133.0	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TONSL	142.9	100.0%	99.7%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TP63	169.9	100.0%	100.0%	Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285
TRAF3IP1	87.5	99.1%	96.7%	Senior-Loken syndrome 9, 616629
TRAIP	128.8	100.0%	100.0%	Seckel syndrome 9, 616777
TRAPPC2	59.0	84.8%	64.8%	Spondyloepiphyseal dysplasia tarda, 313400
TREM2	135.8	100.0%	100.0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TRIP11	87.5	97.2%	91.6%	Osteochondrodysplasia, 184260 Achondrogenesis, type IA, 200600
TRPS1	160.6	100.0%	99.9%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV4	150.4	100.0%	100.0%	Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Scapuloperoneal spinal muscular atrophy, 181405 Metatropic dysplasia, 156530 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Brachyolmia type 3, 113500 ?Avascular necrosis of femoral head, primary, 2, 617383
TRPV6	156.9	100.0%	100.0%	Hyperparathyroidism, transient neonatal, 618188
TTC21B	115.1	99.9%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TWIST1	185.6	100.0%	100.0%	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100

				Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TYROBP	94.9	100.0%	100.0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
UFSP2	126.6	100.0%	99.5%	?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 ?Hip dysplasia, Beukes type, 142669
VAC14	107.4	100.0%	98.8%	Striatonigral degeneration, childhood-onset, 617054
VDR	116.5	99.0%	96.4%	Rickets, vitamin D-resistant, type IIA, 277440
VPS33A	106.3	96.6%	94.7%	Mucopolysaccharidosis-plus syndrome, 617303
WDR19	125.3	100.0%	99.4%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	129.6	100.0%	100.0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	137.8	99.5%	98.3%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	111.7	99.8%	97.8%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WISP3	118.0	100.0%	100.0%	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 Arthropathy, progressive pseudorheumatoid, of childhood, 208230
WNT1	289.4	100.0%	100.0%	Osteogenesis imperfecta, type XV, 615220
WNT10B	176.5	100.0%	100.0%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT3	186.5	100.0%	99.9%	?Tetra-amelia syndrome 1, 273395
WNT5A	174.9	100.0%	100.0%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	136.0	100.0%	100.0%	No OMIM Disease ID
WNT7A	218.7	100.0%	100.0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
XRCC4	139.7	100.0%	99.2%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	138.1	100.0%	99.4%	Desbuquois dysplasia 2, 615777
XYLT2	161.8	99.9%	98.7%	Spondyloocular syndrome, 605822
ZBTB16	161.1	100.0%	100.0%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZMPSTE24	126.2	100.0%	99.7%	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZSWIM6	127.2	97.5%	95.6%	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors