SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS DG-4.1.0 (641 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCC9	96%	96%	100%	99.6%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ACAN	99.1%	98.9%	98.4%	95.4%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361;Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800;Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACP5	100%	100%	100%	98.7%	Spondyloenchondrodysplasia with immune dysregulation, 607944

ACTB	100%	100%	100%	99.3%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ACTG1	100%	100%	100%	99.3%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ACVR1	100%	100%	100%	99.6%	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	100%	100%	100%	98.7%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS15	100%	100%	100%	98.4%	Arthrogryposis, distal, type 12, 620545
ADAMTS17	100%	99.9%	100%	97.2%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	100%	99.8%	100%	99.2%	Geleophysic dysplasia 1, 231050
AFF3	100%	100%	100%	98.7%	KINSSHIP syndrome, 619297
AGA	100%	100%	100%	99.4%	Aspartylglucosaminuria, 208400

AGPS	97.3%	97.3%	100%	99.4%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AIFM1	100%	99.7%	99%	73.5%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
ALG12	100%	100%	100%	98.6%	Congenital disorder of glycosylation, type Ig, 607143
ALG3	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type Id, 601110
ALG9	100%	100%	100%	99.6%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALMS1	100%	100%	100%	99.7%	Alstrom syndrome, 203800
ALPL	100%	100%	100%	98.8%	Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300

ALX1	100%	100%	100%	99.5%	Frontonasal dysplasia 3, 613456
ALX3	100%	99.9%	100%	97.6%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	100%	98.6%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMER1	100%	99.9%	98.9%	69.7%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	99.4%	96.6%	98.7%	70.5%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANAPC1	100%	100%	100%	99.4%	Rothmund-Thomson syndrome, type 1, 618625
ANKH	100%	100%	100%	99%	Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000
ANKRD11	100%	100%	100%	98.7%	KBG syndrome, 148050
ANO5	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260
ANTXR2	96.3%	96.3%	100%	99.4%	Hyaline fibromatosis syndrome, 228600

APC2	100%	100%	100%	98.1%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
ARCN1	100%	100%	100%	99.7%	Short stature-micrognathia syndrome, 617164
ARHGAP31	100%	100%	100%	99.3%	Adams-Oliver syndrome 1, 100300
ARID1A	100%	100%	99.9%	95.6%	Coffin-Siris syndrome 2, 614607
ARID1B	98.5%	98.2%	99.5%	90.8%	Coffin-Siris syndrome 1, 135900
ARSB	100%	100%	100%	99.2%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSK	100%	100%	100%	99.5%	Mucopolysaccharidosis, type X, 619698
ARSL	100%	99.8%	98.5%	69.9%	Chondrodysplasia punctata, X-linked recessive, 302950
ASXL1	100%	100%	100%	99.5%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ATP6V0A2	100%	100%	100%	99.5%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200

ATR	100%	100%	99.7%	98.7%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN1	100%	100%	100%	99.5%	Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864
B3GALT6	96.8%	88.3%	100%	95.4%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465
B3GAT3	96.6%	93.8%	100%	99.2%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	100%	99.9%	100%	99.6%	Peters-plus syndrome, 261540
B4GALT7	100%	100%	100%	99%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BANF1	100%	100%	100%	97.6%	Nestor-Guillermo progeria syndrome, 614008

BGN	100%	99.7%	98.9%	68.6%	Meester-Loeys syndrome, 300989;Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	100%	100%	100%	98.5%	?Camptosynpolydactyly, complex, 607539;Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BLM	96.8%	96.6%	100%	99.7%	Bloom syndrome, 210900
BMP1	100%	100%	100%	99%	Osteogenesis imperfecta, type XIII, 614856
BMP2	100%	100%	100%	98.1%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200
BMP5	100%	99.9%	100%	99.5%	
BMPER	100%	100%	100%	99.7%	Diaphanospondylodysostosis, 608022
BMPR1B	100%	100%	100%	99.9%	Acromesomelic dysplasia 3, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849
BPNT2	100%	100%	100%	98.8%	Chondrodysplasia with joint dislocations, GPAPP type, 614078

BRAF	100%	100%	99.1%	96.2%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
BRF1	100%	100%	100%	99.1%	Cerebellofaciodental syndrome, 616202
BTK	100%	99.7%	99%	71.9%	Agammaglobulinemia, X-linked 1, 300755;Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	100%	100%	100%	99.4%	
BUB1B	100%	100%	100%	99.9%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
CA2	100%	100%	100%	99.8%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730

CANT1	100%	100%	100%	99.2%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719
CASR	100%	100%	100%	99.4%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CBFB	100%	100%	100%	99%	Cleidocranial dysplasia 2, 620099
CBL	100%	100%	100%	99%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CC2D2A	98.2%	98.2%	100%	99.6%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCDC134	100%	100%	100%	99.5%	Osteogenesis imperfecta, type XXII, 619795
CCDC8	99.8%	97.2%	100%	98.6%	3-M syndrome 3, 614205

CCN2	100%	100%	100%	98.8%	
CCN6	100%	100%	100%	99.6%	Progressive pseudorheumatoid dysplasia, 208230
CCNQ	100%	99.7%	98.4%	70.2%	STAR syndrome, 300707
CDC42	100%	100%	100%	99.8%	Takenouchi-Kosaki syndrome, 616737
CDC45	100%	100%	100%	99.2%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	100%	99.1%	?Meier-Gorlin syndrome 5, 613805
CDC73	100%	100%	100%	99.5%	Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001
CDH3	100%	100%	100%	99.2%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDK10	100%	100%	100%	98.9%	Al Kaissi syndrome, 617694

CDKN1C	100%	100%	100%	97%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CDT1	100%	99.9%	100%	98.9%	Meier-Gorlin syndrome 4, 613804
CENPE	100%	100%	100%	99.4%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPJ	100%	100%	100%	99.5%	
CEP120	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP152	100%	100%	100%	99.9%	Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823
CEP290	100%	100%	100%	99.6%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP57	100%	100%	100%	99.7%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	92.8%	92.8%	100%	99.7%	?Seckel syndrome 6, 614728

CFAP410	100%	100%	100%	98.5%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CHST11	100%	100%	100%	99.2%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	100%	100%	100%	97.1%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	100%	99.9%	97.8%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHSY1	99.8%	99%	100%	97.9%	Temtamy preaxial brachydactyly syndrome, 605282
CILK1	100%	100%	100%	99.7%	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924;Endocrine-cerebroosteodysplasia, 612651
CKAP2L	100%	100%	100%	99.7%	Filippi syndrome, 272440
CLCN5	100%	99.7%	99.1%	73.4%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease 1, 300009;Nephrolithiasis, type I, 310468

CLCN7	100%	100%	100%	99%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
COG1	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG4	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COL10A1	100%	100%	100%	99.3%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	100%	100%	100%	99.6%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932

COL11A2	100%	100%	100%	98.2%	Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL1A1	100%	100%	100%	98.8%	Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420

COL1A2	100%	100%	100%	99.8%	Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210
COL27A1	100%	100%	100%	98.9%	Steel syndrome, 615155

COL2A1	100%	100%	100%	99.5%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450;SED congenita, 183900;Kniest dysplasia, 156550;Stickler syndrome, type I, nonsyndromic ocular, 609508;Osteoarthritis with mild chondrodysplasia, 604864;Stickler syndrome, type I, 108300;Platyspondylic skeletal dysplasia, Torrance type, 151210;Spondyloepiphyseal dysplasia, Stanescu type, 616583;Avascular necrosis of the femoral head, 608805;Legg-Calve-Perthes disease, 150600
COL9A1	100%	100%	100%	99.4%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135

COL9A2	100%	100%	100%	98.4%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284
COL9A3	100%	100%	100%	98.4%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
COLEC10	100%	100%	100%	99.7%	3MC syndrome 3, 248340
COLEC11	100%	100%	100%	99%	3MC syndrome 2, 265050
COMP	100%	100%	100%	98.1%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
CPLANE1	100%	100%	100%	99.5%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CREB3L1	100%	100%	100%	97.1%	Osteogenesis imperfecta, type XVI, 616229
CREBBP	100%	100%	100%	99%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CRIPT	100%	100%	100%	99.4%	Rothmund-Thomson syndrome, type 3, 615789
CRTAP	100%	100%	100%	99%	Osteogenesis imperfecta, type VII, 610682

CSF1R	100%	100%	100%	99.2%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSGALNACT1	100%	100%	100%	99.2%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CSPP1	96.9%	96.9%	100%	99.4%	Joubert syndrome 21, 615636
CTSA	100%	99.9%	100%	99.2%	Galactosialidosis, 256540
CTSK	100%	100%	100%	99.2%	Pycnodysostosis, 265800
CUL7	100%	100%	100%	98.6%	3-M syndrome 1, 273750
CYP26B1	100%	100%	100%	98.7%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	100%	100%	100%	99.4%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	100%	100%	100%	99.5%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
DDR1	100%	100%	100%	99.2%	
DDR2	100%	100%	100%	99.6%	Warburg-Cinotti syndrome, 618175;Spondylometaepiphyseal dysplasia, short limb-hand type, 271665

DDRGK1	100%	100%	100%	98.8%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DHCR24	100%	100%	100%	98.8%	Desmosterolosis, 602398
DHODH	100%	100%	100%	98.9%	Miller syndrome, 263750
DLL3	100%	100%	100%	97.8%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100%	100%	100%	98.6%	Adams-Oliver syndrome 6, 616589
DLX3	100%	100%	100%	97.7%	Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510
DLX5	100%	100%	100%	98.8%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	100%	100%	98.2%	87%	
DMP1	100%	100%	100%	98.8%	Hypophosphatemic rickets, AR, 241520
DNA2	100%	100%	100%	99.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807

DNAJC21	100%	100%	100%	98.4%	Bone marrow failure syndrome 3, 617052
DNMT3A	100%	100%	100%	98.9%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
DOCK6	100%	100%	100%	98.8%	Adams-Oliver syndrome 2, 614219
DONSON	100%	100%	100%	99.8%	Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230
DPCD	100%	100%	100%	98.3%	
DPF2	95.7%	95.7%	100%	99.3%	Coffin-Siris syndrome 7, 618027
DPM1	98.3%	93.5%	97.2%	90.4%	Congenital disorder of glycosylation, type Ie, 608799
DSE	100%	100%	100%	99.8%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DVL1	100%	100%	100%	99.2%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100%	100%	100%	98.3%	Robinow syndrome, autosomal dominant 3, 616894

DYM	100%	100%	100%	99.8%	Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800
DYNC2H1	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	100%	100%	100%	99.5%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DYNC2I2	100%	100%	100%	98.5%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYNLT2B	100%	100%	100%	98.8%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EBP	100%	99.9%	98.7%	68.3%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECEL1	100%	100%	100%	97.4%	Arthrogryposis, distal, type 5D, 615065
EDN1	100%	100%	100%	99.8%	Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706
EDNRA	100%	100%	100%	99.6%	{Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367

EFL1	99.2%	99.2%	100%	99.8%	Shwachman-Diamond syndrome 2, 617941
EFNB1	100%	99.8%	97.5%	67.7%	Craniofrontonasal dysplasia, 304110
EFTUD2	100%	100%	100%	99.1%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2AK3	100%	100%	100%	99.7%	Wolcott-Rallison syndrome, 226980
EIF4A3	100%	100%	100%	99.1%	Robin sequence with cleft mandible and limb anomalies, 268305
ELMO2	100%	100%	100%	99.8%	Vascular malformation, primary intraosseous, 606893
EN1	100%	100%	100%	95.8%	?ENDOVE syndrome, limb-brain type, 619218
ENPP1	99.9%	99.3%	100%	99.6%	{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522
EOGT	97.8%	93.7%	100%	99.8%	Adams-Oliver syndrome 4, 615297

EP300	100%	100%	100%	99.2%	Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684
ERCC4	100%	100%	100%	99.5%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERF	100%	100%	99.9%	94.5%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180
ERI1	100%	100%	100%	99.7%	Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663
ESCO2	100%	100%	100%	99.7%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300
EVC	100%	99.8%	100%	99.3%	Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530
EVC2	100%	100%	100%	99.1%	Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530

EXOC6B	100%	100%	100%	99.3%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC5	100%	100%	100%	97%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXT1	100%	100%	100%	99.5%	Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300
EXT2	100%	100%	100%	99.7%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701
EXTL3	100%	99.5%	100%	99.7%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	100%	100%	100%	99.5%	Weaver syndrome, 277590
FAM111A	100%	100%	100%	99.9%	Kenny-Caffey syndrome, type 2, 127000;Gracile bone dysplasia, 602361
FAM20B	100%	100%	100%	99.9%	
FAM20C	100%	100%	100%	97.4%	Raine syndrome, 259775
FANCA	100%	100%	100%	99.4%	Fanconi anemia, complementation group A, 227650
FANCB	96.2%	95.9%	99.1%	73.4%	Fanconi anemia, complementation group B, 300514

FANCC	100%	100%	100%	99.6%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	100%	99.6%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	100%	98.3%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	99.4%	Fanconi anemia, complementation group F, 603467
FANCG	100%	100%	100%	98.9%	Fanconi anemia, complementation group G, 614082
FANCI	100%	100%	100%	99.6%	Fanconi anemia, complementation group I, 609053
FANCL	91%	88.1%	99.8%	97.1%	Fanconi anemia, complementation group L, 614083
FAR1	100%	100%	100%	99.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FBLN1	100%	100%	100%	98.6%	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180

FBN1	100%	100%	100%	99.8%	Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900
FBN2	99.2%	99.2%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBXW4	100%	100%	100%	98%	
FERMT3	100%	100%	100%	99.3%	Leukocyte adhesion deficiency, type III, 612840
FGD1	100%	98.9%	98.7%	67.8%	Intellectual developmental disorder, X-linked syndromic 16, 305400;Aarskog-Scott syndrome, 305400
FGF10	100%	100%	100%	100%	LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920
FGF16	100%	99.3%	97.8%	69.3%	Metacarpal 4-5 fusion, 309630

FGF23	100%	100%	100%	98.6%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100
FGF8	100%	100%	100%	97.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	100%	99.9%	Multiple synostoses syndrome 3, 612961
FGFR1	99.8%	98.9%	100%	99.1%	Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001

FGFR2	100%	100%	100%	99.7%	Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400

FGFR3	100%	100%	100%	99.2%	Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
FIG4	98.4%	98.4%	100%	99.6%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FKBP10	100%	100%	100%	98.9%	Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450

FKBP14	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLNA	100%	99.8%	98.2%	67.3%	Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620
FLNB	100%	100%	100%	99.2%	Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310
FMN1	100%	100%	100%	98.5%	

FN1	100%	100%	100%	99.5%	Spondylometaphyseal dysplasia, corner fracture type, 184255;Glomerulopathy with fibronectin deposits 2, 601894
FUCA1	100%	100%	100%	99%	Fucosidosis, 230000
FUZ	100%	100%	100%	99.1%	{Neural tube defects, susceptibility to}, 182940
FZD2	100%	100%	99.6%	94%	Omodysplasia 2, 164745
GALNS	100%	100%	100%	98.3%	Mucopolysaccharidosis IVA, 253000
GALNT2	100%	100%	100%	98.7%	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	100%	100%	100%	99.9%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GBA1	100%	100%	100%	99.1%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600

GCM2	100%	100%	100%	99.6%	Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343
GDF3	100%	100%	100%	99.6%	Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704
GDF5	100%	100%	100%	99%	Acromesomelic dysplasia 2A, 200700;Acromesomelic dysplasia 2B, 228900;Multiple synostoses syndrome 2, 610017;Symphalangism, proximal, 1B, 615298;Brachydactyly, type A2, 112600;?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072

GDF6	100%	100%	100%	98.1%	Microphthalmia with coloboma 6, digenic, 613703;Microphthalmia, isolated 4, 613094;Leber congenital amaurosis 17, 615360;Multiple synostoses syndrome 4, 617898;Klippel-Feil syndrome 1, autosomal dominant, 118100
GH1	100%	100%	100%	99%	Kowarski syndrome, 262650;Growth hormone deficiency, isolated, type II, 173100;Growth hormone deficiency, isolated, type IB, 612781;Growth hormone deficiency, isolated, type IA, 262400
GHR	99.8%	99.8%	99.8%	98.2%	Laron dwarfism, 262500;Increased responsiveness to growth hormone, 604271;Growth hormone insensitivity, partial, 604271;{Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	100%	100%	100%	99.3%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	100%	100%	100%	98.7%	Growth hormone deficiency, isolated partial, 615925
GINS2	100%	100%	100%	99.1%	

GJA1	100%	100%	100%	99.1%	Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850
GLB1	100%	100%	100%	99.5%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLI1	100%	100%	100%	98.9%	Polydactyly, preaxial I, 174400;Polydactyly, postaxial, type A8, 618123
GLI2	100%	100%	100%	98.7%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829

GLI3	99.3%	99.3%	100%	99.1%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GMNN	100%	100%	100%	99.9%	Meier-Gorlin syndrome 6, 616835
GNAI3	100%	100%	100%	99.4%	Auriculocondylar syndrome 1, 602483
GNAS	99.9%	98.3%	99.9%	95.6%	Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;McCune-Albright syndrome, somatic mosaic, 174800;Pseudohypoparathyroidism Ib, 603233;Pseudopseudohypoparathyroidism, 612463;ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080
GNPAT	100%	100%	100%	99.7%	Rhizomelic chondrodysplasia punctata, type 2, 222765

GNPNAT1	100%	100%	100%	99.6%	?Rhizomelic dysplasia, Ain-Naz type, 619598
GNPTAB	100%	100%	100%	99.5%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	100%	100%	100%	99.2%	Mucolipidosis III gamma, 252605
GNS	100%	100%	100%	99.6%	Mucopolysaccharidosis type IIID, 252940
GORAB	100%	100%	100%	99%	Geroderma osteodysplasticum, 231070
GPC3	100%	99.5%	98.9%	71.1%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	100%	99.8%	98.9%	72.4%	Keipert syndrome, 301026
GPC6	100%	100%	100%	99.4%	Omodysplasia 1, 258315
GPR161	100%	100%	100%	99.3%	{Medulloblastoma predisposition syndrome}, 155255
GPX4	100%	99.8%	100%	99.3%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GSC	100%	100%	100%	97.3%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GUSB	100%	100%	100%	98.7%	Mucopolysaccharidosis VII, 253220

GZF1	100%	100%	100%	99.4%	Joint laxity, short stature, and myopia, 617662
H19					
HAAO	100%	100%	100%	99.6%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HDAC4	100%	100%	100%	98.9%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC8	97.6%	96.8%	98.6%	71.6%	Cornelia de Lange syndrome 5, 300882
HES7	100%	100%	100%	97.2%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	100%	100%	100%	99.5%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	92.4%	92.4%	100%	99.3%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HHAT	100%	100%	100%	99.3%	Nivelon-Nivelon-Mabille syndrome, 600092
HMGA2	94.4%	83.5%	100%	99.3%	Silver-Russell syndrome 5, 618908

HOXA11	100%	100%	100%	97.5%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	99%	95.2%	95.3%	70.6%	Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305
HOXD13	100%	100%	100%	96.1%	Syndactyly, type V, 186300;Synpolydactyly 1, 186000;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200;?Brachydactyly-syndactyly syndrome, 610713
HPGD	100%	100%	100%	99.1%	?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100

HRAS	100%	100%	100%	98.6%	Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040
HS2ST1	100%	100%	100%	99.6%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HSPA9	100%	100%	100%	99.6%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPG2	100%	100%	100%	99.1%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800
HYLS1	100%	100%	100%	99.6%	Hydrolethalus syndrome, 236680
IARS2	100%	100%	100%	99.5%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007

ID4	100%	99.9%	99.9%	92.4%	
IDH1	100%	100%	100%	99.9%	{Glioma, susceptibility to, somatic}, 137800
IDH2	100%	100%	100%	98.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDS	100%	99.3%	99.2%	72.1%	Mucopolysaccharidosis II, 309900
IDUA	100%	100%	100%	97.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IFIH1	100%	100%	100%	99.8%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFITM5	100%	100%	100%	99.4%	Osteogenesis imperfecta, type V, 610967
IFT122	100%	100%	100%	99.3%	Cranioectodermal dysplasia 1, 218330
IFT140	100%	100%	100%	99%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781

IFT172	100%	100%	100%	99.3%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100%	100%	100%	99.4%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	94.9%	94.9%	100%	99.9%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100%	100%	100%	99.6%	Insulin-like growth factor I deficiency, 608747
IGF1R	100%	100%	100%	99.3%	Insulin-like growth factor I, resistance to, 270450
IGF2	100%	100%	100%	99.1%	Silver-Russell syndrome 3, 616489
IGFALS	100%	100%	100%	99.1%	Acid-labile subunit, deficiency of, 615961

IGSF1	100%	99.6%	99.2%	72.2%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	100%	100%	100%	98.7%	Acrocapitofemoral dysplasia, 607778;Brachydactyly, type A1, 112500
IKBKB	100%	100%	100%	99.3%	Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204
IKBKG	96.4%	94.1%	99.3%	75%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081
IL1RN	100%	100%	100%	99.8%	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852

IL2RG	100%	99.8%	98.2%	68.6%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL6ST	100%	100%	100%	99.6%	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752;Stuve-Wiedemann syndrome 2, 619751;Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523;?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750
INPPL1	100%	100%	100%	98.8%	Opsismodysplasia, 258480
INTU	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925
JAG1	100%	100%	100%	99.4%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500

KAT6B	100%	100%	100%	99.4%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KCNJ2	100%	100%	100%	99.2%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KDELR2	100%	100%	100%	99%	Osteogenesis imperfecta, type XXI, 619131
KIAA0586	95.6%	95.6%	100%	99.8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	100%	100%	100%	99.6%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA0825	100%	100%	100%	99.8%	Polydactyly, postaxial, type A10, 618498
KIF22	100%	100%	100%	98.9%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF24	100%	100%	100%	99.6%	
KIF5B	100%	100%	100%	99.8%	

KIF7	100%	99.8%	100%	98.4%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
KL	99.7%	99.1%	99.6%	97.4%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KMT2A	99.2%	99.2%	100%	99.2%	Wiedemann-Steiner syndrome, 605130

KRAS	100%	100%	100%	99.8%	Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800
KYNU	100%	100%	100%	99.6%	?Hydroxykynureninuria, 236800;Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
LAMA5	100%	100%	100%	98.7%	Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076

LBR	100%	100%	100%	99.5%	Pelger-Huet anomaly, 169400;?Reynolds syndrome, 613471;Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019;Greenberg skeletal dysplasia, 215140
LBX1	100%	100%	100%	98.4%	?Central hypoventilation syndrome, congenital, 3, 619483
LEMD3	100%	100%	100%	98.7%	Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700
LFNG	97.8%	92.4%	99.8%	94.9%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	100%	100%	100%	99.1%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100%	100%	100%	99.1%	Pituitary hormone deficiency, combined, 4, 262700
LIFR	100%	100%	100%	99.9%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559

LMBR1	100%	100%	100%	99.8%	Syndactyly, type IV, 186200;Laurin-Sandrow syndrome, 135750;Acheiropody, 200500;Triphalangeal thumb-polysyndactyly syndrome, 190605
LMNA	100%	100%	100%	98.7%	Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112
LMX1B	100%	100%	100%	97.8%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LONP1	100%	100%	100%	98.9%	CODAS syndrome, 600373

LPIN2	99.5%	99.2%	100%	99.5%	Majeed syndrome, 609628
LRP4	100%	100%	100%	99.3%	?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780
LRP5	100%	100%	100%	98.3%	Osteopetrosis, autosomal dominant 1, 607634;[Bone mineral density variability 1], 601884;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;Exudative vitreoretinopathy 4, 601813
LRRK1	100%	100%	100%	99.5%	Osteosclerotic metaphyseal dysplasia, 615198
LTBP1	100%	100%	100%	99.1%	Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	100%	100%	100%	99%	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	100%	100%	100%	98.1%	Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
LYSET	100%	100%	100%	99.8%	Dysostosis multiplex, Ain-Naz type, 619345
LZTR1	100%	100%	100%	99.1%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAB21L2	100%	100%	100%	98.8%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAD2L2	100%	100%	100%	99.1%	?Fanconi anemia, complementation group V, 617243
MAFB	100%	100%	100%	98.5%	Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300
MAN2B1	100%	100%	100%	98.3%	Mannosidosis, alpha-, types I and II, 248500
MANBA	100%	100%	100%	99.7%	Mannosidosis, beta, 248510
MAP2K1	95.8%	95.8%	100%	99.5%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100%	100%	100%	98.2%	Cardiofaciocutaneous syndrome 4, 615280

MAP3K20	100%	100%	100%	99.4%	Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	100%	99.9%	100%	99.5%	Frontometaphyseal dysplasia 2, 617137;Cardiospondylocarpofacial syndrome, 157800
MAPK1	100%	100%	100%	99.1%	Noonan syndrome 13, 619087
MAPKAPK5	100%	100%	100%	99.6%	Neurocardiofaciodigital syndrome, 619869
MASP1	100%	100%	100%	99.4%	3MC syndrome 1, 257920
MATN3	100%	100%	100%	99.1%	{Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728;Epiphyseal dysplasia, multiple, 5, 607078
MBTPS1	99.8%	99.2%	100%	99.3%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392

MBTPS2	100%	99.7%	98.9%	75.5%	Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918
MCM5	100%	100%	100%	98.7%	?Meier-Gorlin syndrome 8, 617564
MECOM	100%	100%	100%	99.6%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MEGF8	100%	100%	100%	98.5%	Carpenter syndrome 2, 614976
MEOX1	100%	100%	100%	98.6%	Klippel-Feil syndrome 2, 214300
MESD	100%	100%	100%	98.4%	Osteogenesis imperfecta, type XX, 618644
MESP2	99.9%	98.1%	100%	98.7%	Spondylocostal dysostosis 2, autosomal recessive, 608681

MET	100%	100%	100%	99.8%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705
MGP	100%	100%	100%	99.1%	Keutel syndrome, 245150
MIR140					Spondyloepiphyseal dysplasia, Nishimura type, 618618
MKS1	99.1%	99%	100%	99.2%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MMP13	92.1%	92.1%	100%	99.5%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111;Metaphyseal dysplasia, Spahr type, 250400
MMP14	94.9%	94.9%	100%	98.4%	Winchester syndrome, 277950
MMP2	100%	100%	100%	98.6%	Multicentric osteolysis, nodulosis, and arthropathy, 259600

MMP9	100%	100%	100%	98.1%	Metaphyseal anadysplasia 2, 613073
MNX1	92.5%	86.3%	96.9%	79.1%	Currarino syndrome, 176450
MRAS	100%	100%	100%	99.6%	Noonan syndrome 11, 618499
MSX2	100%	100%	100%	99%	Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500
MTAP	100%	100%	100%	98.8%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTX2	100%	100%	100%	99.5%	Mandibuloacral dysplasia progeroid syndrome, 619127
MYCN	100%	100%	100%	97.1%	Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748

MYH3	100%	100%	100%	99.3%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469;Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436;Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYL11	100%	100%	100%	98.9%	Arthrogryposis, distal, type 1C, 619110
MYO18B	100%	100%	100%	98.8%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
NADSYN1	100%	100%	100%	99.2%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGLU	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NANS	100%	100%	100%	99.1%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442

NBAS	100%	99.9%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NEK1	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK9	100%	100%	100%	99.5%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022
NEPRO	100%	100%	100%	99.8%	Anauxetic dysplasia 3, 618853
NEU1	100%	100%	100%	99.3%	Sialidosis, type II, 256550;Sialidosis, type I, 256550

NF1	99.4%	99.4%	100%	99.7%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFIX	99.8%	98.8%	99.9%	96.8%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NIN	100%	100%	100%	99.5%	?Seckel syndrome 7, 614851
NIPBL	100%	100%	100%	99.7%	Cornelia de Lange syndrome 1, 122470
NKX3-2	100%	100%	100%	99.1%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP3	100%	100%	100%	99.3%	CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendothelitis fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900

NMNAT1	100%	97.8%	100%	99.8%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553
NOG	100%	100%	100%	97.5%	Symphalangism, proximal, 1A, 185800;Brachydactyly, type B2, 611377;Stapes ankylosis with broad thumbs and toes, 184460;Tarsal-carpal coalition syndrome, 186570;Multiple synostoses syndrome 1, 186500
NOTCH1	99.4%	99.1%	100%	98.8%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	100%	100%	100%	99.7%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NPPC	100%	100%	100%	99.7%	
NPR2	100%	99.9%	100%	99%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100%	100%	100%	98.5%	Boudin-Mortier syndrome, 619543

NRAS	100%	100%	100%	99.6%	Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
NSD1	100%	100%	100%	99.5%	Sotos syndrome, 117550
NSD2	99.5%	99.4%	100%	98.9%	Rauch-Steindl syndrome, 619695
NSDHL	100%	99.3%	99%	72.3%	CK syndrome, 300831;CHILD syndrome, 308050
NSMCE2	100%	100%	100%	99.4%	Seckel syndrome 10, 617253
NXN	100%	100%	100%	98.4%	Robinow syndrome, autosomal recessive 2, 618529
OBSL1	100%	100%	100%	98.7%	3-M syndrome 2, 612921

OFD1	100%	99.9%	99%	74.1%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
ORC1	100%	100%	100%	99.4%	Meier-Gorlin syndrome 1, 224690
ORC4	100%	100%	100%	99.9%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	100%	100%	99.7%	Meier-Gorlin syndrome 3, 613803
OSTM1	99.8%	98.5%	100%	99.5%	Osteopetrosis, autosomal recessive 5, 259720
OTX2	100%	100%	100%	99.7%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
P3H1	100%	100%	100%	99.3%	Osteogenesis imperfecta, type VIII, 610915
P4HB	96%	94.6%	100%	99%	Cole-Carpenter syndrome 1, 112240
PAM16	84.5%	84.5%	100%	99%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	100%	100%	100%	99.4%	Short stature, Dauber-Argente type, 619489

PAPSS2	100%	100%	100%	99.3%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PAX3	100%	100%	100%	99.1%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PCNT	100%	100%	100%	99.2%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	100%	100%	100%	99.6%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680
PDE3A	100%	100%	100%	99.3%	Hypertension and brachydactyly syndrome, 112410
PDE4D	100%	100%	100%	99.2%	Acrodysostosis 2, with or without hormone resistance, 614613

PDGFRB	100%	100%	100%	99%	Premature aging syndrome, Penttinen type, 601812;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007;Myeloproliferative disorder with eosinophilia, 131440
PEX5	100%	100%	100%	98.5%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	98.5%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	97.9%	97.9%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PHEX	100%	99.8%	99.3%	75.2%	Hypophosphatemic rickets, X-linked dominant, 307800

PHGDH	100%	100%	100%	99.4%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PIGV	100%	99.8%	100%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PIK3R1	100%	99.9%	100%	99.9%	Immunodeficiency 36, 616005;?Agammaglobulinemia 7, autosomal recessive, 615214;SHORT syndrome, 269880
PISD	100%	100%	100%	99.4%	Liberfarb syndrome, 618889
PITX1	100%	100%	100%	95.8%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	100%	100%	100%	97.9%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PKDCC	100%	100%	99.7%	88.9%	Rhizomelic limb shortening with dysmorphic features, 618821
PLAG1	100%	100%	100%	99.7%	Adenomas, salivary gland pleomorphic, somatic, 181030;Silver-Russell syndrome 4, 618907

PLCB3	100%	100%	100%	98.9%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	99%	99%	100%	99.8%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
PLEKHM1	100%	100%	100%	98.6%	?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107
PLK4	100%	100%	100%	99.5%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100%	100%	100%	98.4%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100%	100%	100%	99.7%	Bruck syndrome 2, 609220
PLOD3	100%	100%	100%	98.4%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLS3	96.8%	96.6%	99%	73.5%	Bone mineral density QTL18, osteoporosis, 300910;Diaphragmatic hernia 5, X-linked, 306950

PNPLA6	100%	100%	100%	98.8%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
POC1A	100%	100%	100%	99.4%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POLE	100%	100%	100%	99%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
POLL	100%	100%	100%	99.4%	
POLR1A	100%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462
POLR1B	100%	100%	100%	99.8%	Treacher-Collins syndrome 4, 618939
POLR1C	83.5%	83.2%	100%	98.8%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	100%	99.8%	Treacher Collins syndrome 2, 613717

POLR3A	100%	100%	100%	99.3%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3GL	100%	100%	100%	99.4%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POP1	100%	100%	100%	99.6%	Anauxetic dysplasia 2, 617396
POR	100%	100%	100%	99.3%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	100%	99.5%	98.3%	69.5%	Focal dermal hypoplasia, 305600
POU1F1	100%	100%	100%	99.6%	Pituitary hormone deficiency, combined or isolated, 1, 613038

PPIB	100%	100%	100%	98.5%	Osteogenesis imperfecta, type IX, 259440
PPM1D	100%	100%	100%	99.5%	Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450
PPP1CB	88%	87.4%	100%	99.6%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R21	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PRG4	100%	100%	100%	94.3%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRKACA	100%	99.9%	100%	99.1%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830;Cardioacrofacial dysplasia 1, 619142
PRKACB	100%	100%	100%	99.5%	Cardioacrofacial dysplasia 2, 619143

PRKAR1A	100%	100%	100%	99.4%	Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960
PRKG2	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia, Pagnamenta type, 619638;Acromesomelic dysplasia 4, 619636
PRMT7	100%	100%	100%	98.6%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PROKR2	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100%	100%	100%	98.8%	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	100%	100%	100%	99.5%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992

PSMB1	100%	100%	100%	99.9%	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038
PTBP1	100%	100%	100%	97.9%	
PTDSS1	100%	100%	100%	99.4%	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	100%	100%	100%	98.9%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045
PTHLH	100%	100%	100%	99.6%	Brachydactyly, type E2, 613382
PTPN11	89.7%	89.2%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PUF60	100%	100%	100%	98.7%	Verheij syndrome, 615583
PYCR1	100%	100%	100%	99.4%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	100%	100%	100%	99.6%	Carpenter syndrome, 201000

RAB33B	100%	100%	100%	99.6%	Smith-McCort dysplasia 2, 615222
RAC3	100%	100%	100%	98.5%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	100%	100%	100%	99.8%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RAD51	89.3%	89.3%	100%	99.4%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244
RAF1	96.6%	93.5%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RALA	100%	100%	100%	100%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RASGRP2	100%	100%	100%	98.6%	?Bleeding disorder, platelet-type, 18, 615888
RBBP8	100%	100%	100%	99.5%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic
RBM8A	100%	100%	100%	99.6%	Thrombocytopenia-absent radius syndrome, 274000

RBPJ	100%	100%	100%	99.8%	Adams-Oliver syndrome 3, 614814
RECQL4	100%	100%	100%	98.9%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
RFWD3	100%	100%	100%	99.8%	?Fanconi anemia, complementation group W, 617784
RIGI	98.6%	98.6%	100%	99.6%	Singleton-Merten syndrome 2, 616298
RIPPLY2	100%	100%	100%	99.1%	?Spondylocostal dysostosis 6, 616566
RIT1	100%	100%	100%	99%	Noonan syndrome 8, 615355
RMRP					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RNPC3	100%	100%	100%	99.8%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU4ATAC					Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710

ROR2	100%	100%	100%	99.4%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPL10	100%	99.3%	98.3%	70.8%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RPL13	100%	99.2%	100%	98.8%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RRAS	100%	99.7%	100%	97.2%	
RRAS2	100%	100%	100%	99.3%	Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	100%	100%	100%	98.3%	
RSPO2	100%	100%	100%	99.9%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022;Tetraamelia syndrome 2, 618021
RSPRY1	100%	100%	100%	99.6%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723

RUNX2	100%	100%	100%	98.4%	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510;Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Cleidocranial dysplasia, 119600
SALL1	100%	100%	100%	99.1%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	100%	100%	100%	99.2%	?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323
SATB2	100%	100%	100%	99.5%	Glass syndrome, 612313
SBDS	100%	100%	100%	99.7%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SCARF2	100%	100%	100%	95.1%	Van den Ende-Gupta syndrome, 600920
SCUBE3	100%	100%	100%	99%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184
SEC24D	100%	100%	100%	99.3%	Cole-Carpenter syndrome 2, 616294

SEMA3A	100%	100%	100%	99.8%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SERPINF1	100%	100%	100%	98.8%	Osteogenesis imperfecta, type VI, 613982
SERPINH1	100%	100%	100%	99.3%	{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848
SETD2	100%	100%	100%	99.5%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SF3B4	100%	100%	100%	98.3%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100%	100%	100%	99.4%	Pyle disease, 265900
SGMS2	100%	100%	100%	99.8%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGSH	100%	100%	100%	99.3%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3BP2	99.9%	99%	100%	98.8%	Cherubism, 118400
SH3PXD2B	100%	100%	100%	98.8%	Frank-ter Haar syndrome, 249420

SHH	100%	100%	100%	97.1%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SHOC2	100%	100%	100%	99.7%	Noonan syndrome-like with loose anagen hair 1, 607721
SHOX	47.4%	47.4%	50%	49.4%	Short stature, idiopathic familial, 300582;Leri-Weill dyschondrosteosis, 127300;Langer mesomelic dysplasia, 249700;Short stature, idiopathic familial, 300582;Langer mesomelic dysplasia, 249700;Leri-Weill dyschondrosteosis, 127300
SKI	100%	99.8%	100%	96.8%	Shprintzen-Goldberg syndrome, 182212
SLC10A7	92.8%	92.8%	100%	99.8%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC17A5	100%	100%	100%	99.9%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC25A24	99.5%	99.5%	99.6%	97.4%	Fontaine progeroid syndrome, 612289

SLC26A2	100%	100%	100%	99.7%	Epiphyseal dysplasia, multiple, 4, 226900;De la Chapelle dysplasia, 256050;Diastrophic dysplasia, 222600;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Achondrogenesis Ib, 600972;Atelosteogenesis, type II, 256050
SLC29A3	100%	100%	100%	99.3%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	100%	100%	100%	97.3%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35C1	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	100%	100%	99.7%	Schneckenbecken dysplasia, 269250
SLC39A13	100%	100%	100%	97.7%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC4A2	100%	100%	100%	98.9%	?Osteopetrosis, autosomal recessive 9, 620366

SLCO2A1	100%	100%	100%	99%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441
SLCO5A1	100%	100%	100%	99.5%	
SLX4	100%	100%	100%	99%	Fanconi anemia, complementation group P, 613951
SMAD2	100%	100%	100%	99.6%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	100%	100%	100%	98.7%	Loeys-Dietz syndrome 3, 613795
SMAD4	100%	100%	100%	99.9%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	100%	100%	100%	96.5%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439

SMARCA4	100%	100%	100%	98.7%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMARCA5	100%	100%	100%	99.2%	
SMARCAL1	100%	100%	100%	99.4%	Schimke immunoosseous dysplasia, 242900
SMARCB1	100%	99.9%	100%	98.1%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCE1	100%	100%	100%	99.5%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SMC1A	100%	99.6%	98.9%	69.4%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	100%	99.7%	Cornelia de Lange syndrome 3, 610759

SMO	100%	100%	100%	99.2%	Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	100%	100%	100%	99.6%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNRPB	100%	100%	100%	99.5%	Cerebrocostomandibular syndrome, 117650
SNX10	89.3%	89.3%	100%	99.8%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	98.7%	98.3%	100%	99.7%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	100%	100%	100%	99.3%	Noonan syndrome 9, 616559
SOST	100%	100%	100%	99.4%	Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX2	100%	99.9%	100%	96.8%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900

SOX3	100%	100%	95.2%	58.4%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX9	100%	100%	100%	96.9%	Campomelic dysplasia with autosomal sex reversal, 114290;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290
SP7	100%	100%	100%	98.9%	Osteogenesis imperfecta, type XII, 613849
SPARC	100%	100%	100%	99.1%	Osteogenesis imperfecta, type XVII, 616507
SPECC1L	100%	99.2%	100%	99.4%	Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251
SPINK5	100%	100%	100%	99.8%	Netherton syndrome, 256500
SPR	100%	100%	100%	98.9%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100%	100%	100%	99.7%	Legius syndrome, 611431
SPRED2	100%	100%	100%	99.3%	Noonan syndrome 14, 619745

SRCAP	100%	100%	100%	98.7%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140
SRP54	100%	100%	100%	99.8%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
STAT3	100%	100%	100%	99.7%	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	100%	100%	100%	99.2%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985
STIM1	100%	99.8%	100%	99.2%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
SULF1	100%	100%	100%	99.6%	
SUMF1	100%	100%	100%	99.5%	Multiple sulfatase deficiency, 272200

TAB2	100%	100%	100%	98.8%	Congenital heart defects, nonsyndromic, 2, 614980
TAPT1	100%	100%	100%	99.4%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBCE	100%	100%	100%	99.4%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBX15	100%	100%	100%	99.4%	Cousin syndrome, 260660
TBX2	99.8%	97.6%	100%	97.7%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX3	100%	100%	100%	97.8%	Ulnar-mammary syndrome, 181450
TBX4	100%	100%	100%	98.7%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891;Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX5	100%	100%	100%	99.1%	Holt-Oram syndrome, 142900

TBX6	100%	100%	100%	98.7%	Spondylocostal dysostosis 5, 122600
TBXAS1	100%	100%	100%	99.4%	Ghosal hematodiaphyseal syndrome, 231095
TCF12	100%	100%	100%	99.8%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCIRG1	100%	100%	100%	99%	Osteopetrosis, autosomal recessive 1, 259700
TCOF1	100%	100%	100%	99.1%	Treacher Collins syndrome 1, 154500
TCTN2	98.5%	98.5%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	99.8%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TENT5A	100%	100%	100%	99.4%	Osteogenesis imperfecta, type XVIII, 617952
TGDS	100%	100%	100%	99.7%	Catel-Manzke syndrome, 616145
TGFB1	100%	99.8%	100%	96.7%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213;Camurati-Engelmann disease, 131300;{Cystic fibrosis lung disease, modifier of}, 219700

TGFB2	100%	100%	100%	99.5%	Loeys-Dietz syndrome 4, 614816
TGFB3	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	100%	99.8%	100%	99.1%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	99.6%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239
THPO	100%	100%	100%	99.6%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
TMCO1	87.7%	87.7%	100%	98.4%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMEM165	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type IIk, 614727

TMEM216	100%	100%	100%	99.9%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM231	93.3%	93.2%	100%	98.9%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM38B	100%	100%	100%	99.9%	Osteogenesis imperfecta, type XIV, 615066
TMEM53	100%	100%	100%	99.1%	Craniotubular dysplasia, Ikegawa type, 619727
TMEM67	96.1%	96.1%	100%	99.8%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TNFRSF11A	99.9%	99.2%	100%	98.3%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810
TNFRSF11B	100%	100%	100%	99.8%	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	100%	100%	100%	98.6%	Osteopetrosis, autosomal recessive 2, 259710
TONSL	100%	100%	100%	99.3%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510

TP63	100%	100%	100%	99.6%	Premature ovarian failure 21, 620311;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292;Hay-Wells syndrome, 106260;Split-hand/foot malformation 4, 605289;Orofacial cleft 8, 618149;Rapp-Hodgkin syndrome, 129400;ADULT syndrome, 103285;Limb-mammary syndrome, 603543
TRAF3IP1	100%	100%	100%	98.7%	Senior-Loken syndrome 9, 616629
TRAF7	100%	100%	100%	98.5%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100%	100%	100%	99.4%	Seckel syndrome 9, 616777
TRAPPC2	100%	99.7%	99.4%	75.5%	Spondyloepiphyseal dysplasia tarda, 313400
TREM2	100%	100%	100%	99.5%	{Alzhieimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TRIM37	98.3%	98.3%	100%	99.4%	Mulibrey nanism, 253250

TRIP11	100%	100%	100%	99.7%	Odontochondrodysplasia 1, 184260;Achondrogenesis, type IA, 200600
TRIP13	100%	100%	100%	99.3%	Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598
TRPS1	100%	100%	100%	99.7%	Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350

TRPV4	100%	100%	100%	98.4%	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;SED, Maroteaux type, 184095;Metatropic dysplasia, 156530;Scapuloperoneal spinal muscular atrophy, 181405;Hereditary motor and sensory neuropathy, type IIc, 606071;?Avascular necrosis of femoral head, primary, 2, 617383;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500
TRPV6	100%	100%	100%	99.3%	Hyperparathyroidism, transient neonatal, 618188
TTC21B	98.3%	97.8%	100%	99.8%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820
TTI2	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 39, 615541

TWIST1	100%	99.9%	100%	98.3%	Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TYROBP	100%	100%	100%	98.9%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
UBA2	100%	100%	100%	99.7%	ACCES syndrome, 619959
UBE2T	100%	100%	100%	99%	Fanconi anemia, complementation group T, 616435
UFSP2	100%	100%	100%	99.6%	?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028
VAC14	100%	100%	100%	99.4%	Striatonigral degeneration, childhood-onset, 617054
VDR	100%	100%	100%	99.2%	Rickets, vitamin D-resistant, type IIA, 277440
VPS33A	89.5%	89.5%	100%	99.4%	Mucopolysaccharidosis-plus syndrome, 617303

VPS35L	100%	100%	100%	99.4%	Ritscher-Schinzel syndrome 3, 619135
WDR19	100%	100%	100%	99.7%	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867
WDR35	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WNT1	100%	100%	100%	98.5%	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220
WNT10B	100%	100%	100%	99.3%	Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300
WNT3	100%	99.8%	100%	99.4%	?Tetra-amelia syndrome 1, 273395
WNT5A	100%	100%	99.9%	96.2%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100%	100%	100%	98.2%	

WNT7A	100%	100%	100%	99.5%	Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820
XRCC4	100%	100%	100%	99.8%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	99.9%	98.8%	100%	98%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	99.7%	97.3%	100%	99.1%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822
ZBTB16	96%	96%	100%	99%	Leukemia, acute promyelocytic, PL2F/RARA type
ZC4H2	100%	99.8%	99.1%	71.6%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	100%	100%	100%	99.5%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210

ZSWIM6	96.6%	93.7%	97.4%	93.2%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671

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.
TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.
TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.
srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023.
This list is accurate for panel version DG 4.0.0

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