SHORT STATURE AND SKELETAL DYSPLASIA GENE PANEL DG 3.1.0 (539 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCC9	100	99,9	100	100	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ACAN	96,6	92,9	98,9	98,7	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	99,8	98,3	100	100	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	99,7	96,1	100	100	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVR1	100	100	100	100	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	99,9	98,5	100	100	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	92,8	89	97,6	95,8	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	97,1	93,3	99,8	99,4	Geleophysic dysplasia 1, 231050
AGA	100	100	100	100	Aspartylglucosaminuria, 208400
AGPS	99,3	95,4	100	99,9	Rhizomelic chondrodysplasia punctata, type 3, 600121
AIFM1	99,9	98,8	100	100	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
ALG12	100	100	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG3	100	99,7	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG9	100	99,7	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
ALPL	100	100	100	100	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300

					Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALX1	99,7	97,1	100	100	Frontonasal dysplasia 3, 613456
ALX3	77,9	73,3	100	100	Frontonasal dysplasia 1, 136760
ALX4	100	99,3	100	100	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMER1	99,9	98,5	100	100	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100	99,1	100	100	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKH	100	100	100	100	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKRD11	96,1	93,5	100	100	KBG syndrome, 148050
ANO5	99,5	97,3	100	100	Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Gnathodiaphyseal dysplasia, 166260
ANTXR2	100	98,2	100	100	Hyaline fibromatosis syndrome, 228600
APC2	97,6	92,7	99,9	99,1	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
ARCN1	97	96,6	96,9	96,6	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARHGAP31	99,9	98,8	100	100	Adams-Oliver syndrome 1, 100300
ARID1B	96,2	95,2	97,9	96,7	Coffin-Siris syndrome 1, 135900
ARSB	97	88,7	100	100	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSL	99	93	100	99,9	Chondrodysplasia punctata, X-linked recessive, 302950
ATP6V0A2	100	99,5	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATR	99,9	99,4	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
B3GALT6	75,7	69,7	89,8	81,6	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	99,9	98,2	94,8	94,8	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	99,8	97,4	99,9	98,6	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BGN	100	100	100	100	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	70,9	50,4	99,8	97,3	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539

BMP1	100	100	100	100	Osteogenesis imperfecta, type XIII, 614856
BMP2	100	100	100	100	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877
BMPER	100	99,8	100	100	Diaphanospondylodysostosis, 608022
BMPR1B	100	99,9	100	100	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
BRAF	91	81,1	100	100	Melanoma, malignant, somatic, 155600 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 114500 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0
BRF1	99,9	98,4	100	100	Cerebellofaciodental syndrome, 616202
BTK	100	99,9	100	99,9	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 Agammaglobulinemia, X-linked 1, 300755
BTRC	97,6	97,3	100	100	No OMIM disease ID
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CANT1	100	99,9	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CASR	100	99,9	100	100	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CBL	97,3	97,1	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CCDC134	100	100	100	100	No OMIM disease ID
CCDC8	100	100	100	100	3-M syndrome 3, 614205
CCN6	84,7	84,6	84,6	84,6	Progressive pseudorheumatoid dysplasia, 208230
CCNQ	83,1	78,5	98,9	94,6	STAR syndrome, 300707
CDC42	97,9	90,9	100	100	Takenouchi-Kosaki syndrome, 616737
CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063

CDC6	100	100	100	100	?Meier-Gorlin syndrome 5, 613805
CDC73	100	99,4	100	100	Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism, familial primary, 145000
CDKN1C	88	77,8	99,3	97,3	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDT1	99,7	97,5	100	99,1	Meier-Gorlin syndrome 4, 613804
CENPE	98,2	92,2	100	100	?Microcephaly 13, primary, autosomal recessive, 616051
CEP120	100	99,5	100	100	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP152	99,7	98,2	100	100	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	96,1	90	100	100	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CFAP410	100	99,3	100	100	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
CHST11	100	100	100	100	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	99,9	98,9	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100	99,4	100	100	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHSY1	97,2	95,7	99,7	98	Temtamy preaxial brachydactyly syndrome, 605282
CILK1	99,9	98,7	100	100	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
CKAP2L	99,7	98,6	100	100	Filippi syndrome, 272440
CLCN5	99,9	98,3	100	100	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
CLCN7	99,7	98,4	100	100	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
COG1	100	100	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG4	100	99,9	100	100	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COL10A1	100	98,4	100	100	Metaphyseal chondrodysplasia, Schmid type, 156500

COL11A1	96,2	92,8	100	100	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520
COL11A2	100	99,7	100	100	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL1A1	99,9	98,6	100	100	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 {Bone mineral density variation QTL, osteoporosis}, 166710 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL1A2	99,4	96,9	100	100	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 {Osteoporosis, postmenopausal}, 166710 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	99,9	99,7	100	100	Steel syndrome, 615155
COL2A1	100	99,7	100	100	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 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	100	99,2	100	100	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,9	99	100	100	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	98,7	95,5	99,7	98,6	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COMP	93,4	92,3	100	100	Epiphyseal dysplasia, multiple, 1, 132400 Carpal tunnel syndrome 2, 619161 Pseudoachondroplasia, 177170
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CREB3L1	100	99,9	100	100	Osteogenesis imperfecta, type XVI, 616229
CREBBP	99,7	98,5	100	100	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRIPT	98,1	93,2	100	100	Short stature with microcephaly and distinctive facies, 615789
CRTAP	99,8	98,8	100	100	Osteogenesis imperfecta, type VII, 610682
CSF1R	99,9	99,3	100	100	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSGALNACT1	100	99,8	100	100	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CTSA	100	100	100	100	Galactosialidosis, 256540
CTSK	100	99,9	100	100	Pycnodysostosis, 265800
CUL7	100	99,3	100	100	3-M syndrome 1, 273750
CYP26B1	100	99,9	100	100	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	99,9	99,3	100	100	Vitamin D-dependent rickets, type I, 264700
CYP2R1	99,4	95,6	100	100	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
DDR2	100	99,9	100	100	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 Warburg-Cinotti syndrome, 618175
DDRGK1	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX58	99,9	99	100	100	Singleton-Merten syndrome 2, 616298
DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHODH	100	100	100	100	Miller syndrome, 263750
DLL3	92,1	87	100	99,1	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100	99,2	100	100	Adams-Oliver syndrome 6, 616589

DLX3	99,9	98,4	100	100	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX5	100	99,9	100	100	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	100	100	100	100	No OMIM disease ID
DMP1	100	99,9	100	100	Hypophosphatemic rickets, AR, 241520
DNA2	99,8	98,3	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAJC21	99,8	98,7	100	100	Bone marrow failure syndrome 3, 617052
DNMT3A	99,8	98,6	100	100	Heyn-Sproul-Jackson syndrome, 618724 Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DOCK6	99,3	98,9	100	100	Adams-Oliver syndrome 2, 614219
DONSON	91,7	85,3	100	100	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPCD	100	100	100	100	No OMIM disease ID
DPM1	98,2	91,3	99,7	97,1	Congenital disorder of glycosylation, type Ie, 608799
DSE	99	96,1	100	100	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DVL1	97,2	95	100	100	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100	100	100	100	Robinow syndrome, autosomal dominant 3, 616894
DYM	97,4	96,5	100	100	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC2H1	98,8	95,5	100	100	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	99,7	97,6	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EBP	99,7	95,8	100	100	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECEL1	95,4	90	100	100	Arthrogryposis, distal, type 5D, 615065
EDN1	100	100	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDNRA	100	100	100	100	{Migraine, resistance to}, 157300 Mandibulofacial dysostosis with alopecia, 616367
EFL1	99,6	98,5	100	100	Shwachman-Diamond syndrome 2, 617941
EFNB1	100	100	100	100	Craniofrontonasal dysplasia, 304110
EFTUD2	100	99,8	100	100	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2AK3	97,2	94,5	100	100	Wolcott-Rallison syndrome, 226980

EIF4A3	100	99,5	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
ENPP1	96,4	91,2	98,7	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853
EOGT	79,4	78,4	91,9	89	Adams-Oliver syndrome 4, 615297
EP300	99,8	99	100	100	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
ERF	99,9	98,5	100	100	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ESCO2	98,7	95,2	100	100	Roberts-SC phocomelia syndrome, 268300
EVC	93,9	88,6	96,9	94,8	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	97,7	96,1	100	100	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC6B	99,1	97,6	100	100	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC5	100	100	100	100	No OMIM disease ID
EXT1	99,9	98,4	100	100	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100	99,3	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	100	99,5	100	100	Weaver syndrome, 277590
FAM111A	99,9	99,3	100	100	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM20B	100	99,9	100	100	No OMIM disease ID
FAM20C	100	100	100	99,8	Raine syndrome, 259775
FAR1	97,6	92,8	100	100	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBLN1	99,7	97,6	100	99,7	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180
FBN1	100	99,9	100	100	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	100	99,9	100	100	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXW4	81,6	79	83,5	80	No OMIM disease ID
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FGD1	97,3	92,8	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	100	99,8	100	100	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	99,6	97,5	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF8	98,2	88,9	100	99,6	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100	100	100	100	Multiple synostoses syndrome 3, 612961
FGFR1	100	99,9	100	100	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	97,7	97,1	100	100	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	99,8	97,7	100	99,8	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	100	99,8	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FKBP10	98,8	97,2	100	100	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	100	99,9	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLNA	100	99,9	100	100	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	99,5	98,8	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FMN1	97,3	96,3	100	100	No OMIM disease ID
FN1	100	99,3	100	100	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FUCA1	100	99,9	100	100	Fucosidosis, 230000
FUZ	100	100	100	100	{Neural tube defects, susceptibility to}, 182940

FZD2	99,9	98,2	100	100	Omodysplasia 2, 164745
GALNS	100	99,8	100	100	Mucopolysaccharidosis IVA, 253000
GALNT3	99,8	99	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GCM2	100	100	100	100	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated 2, 618883
GDF3	100	100	100	100	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702
GDF5	100	100	100	100	?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 {Osteoarthritis-5}, 612400 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017
GDF6	100	99,9	100	99,4	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	100	100	100	100	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	99,6	99,5	99,8	99,8	{Hypercholesterolemia, familial, modifier of}, 143890 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 Growth hormone insensitivity, partial, 604271
GHRHR	96,4	96,1	100	100	Growth hormone deficiency, isolated, type IV, 618157
GHSR	98,5	95,8	100	100	Growth hormone deficiency, isolated partial, 615925
GJA1	100	100	100	100	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	99,9	97,4	100	100	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLI2	99,1	97,4	100	99,8	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	98,5	98	100	100	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GMNN	99,8	97,4	100	100	Meier-Gorlin syndrome 6, 616835
GNAI3	99,3	95,2	100	100	Auriculocondylar syndrome 1, 602483
GNAS	86,9	85,1	82	81,7	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	99,7	97,3	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	68,8	48,4	100	100	No OMIM disease ID
GNPTAB	100	99,9	100	100	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	99,1	94,3	100	99,9	Mucolipidosis III gamma, 252605
GNS	98,4	94,8	100	100	Mucopolysaccharidosis type IIID, 252940
GORAB	100	99,1	100	100	Geroderma osteodysplasticum, 231070
GPC3	99,1	94,7	100	100	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	100	100	100	100	Omodysplasia 1, 258315
GPR161	100	100	100	100	No OMIM disease ID
GPX4	90,5	85,8	98,2	94,9	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GSC	99,2	92,4	100	100	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GUSB	92,9	91,7	100	100	Mucopolysaccharidosis VII, 253220
GZF1	100	99,6	100	100	Joint laxity, short stature, and myopia, 617662
HAAO	100	99,8	100	100	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660

HDAC4	100	99,8	100	100	No OMIM disease ID
HDAC8	86,5	85,1	96,3	94,8	Cornelia de Lange syndrome 5, 300882
HES7	84,4	53,9	100	100	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	99,7	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	86,4	86,3	91,2	89,3	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HMGA2	81,3	76,7	75,1	73,8	Silver-Russell syndrome 5, 618908
HOXA11	97,1	87,5	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	77,7	69	89,7	79,7	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXD13	99,9	98,6	100	100	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 ?Brachydactyly-syndactyly syndrome, 610713 Syndactyly, type V, 186300 Synpolydactyly 1, 186000
HPGD	100	98,9	100	100	Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 ?Digital clubbing, isolated congenital, 119900 Cranioosteoarthropathy, 259100
HRAS	100	100	100	100	Costello syndrome, 218040 Bladder cancer, somatic, 109800 Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550
HSPA9	88,5	84,5	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPG2	99,2	97,7	100	99,9	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IARS2	100	99,9	100	100	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
ID4	87,6	82,5	98,9	93,1	No OMIM disease ID
IDH1	93,3	80,1	100	100	{Glioma, susceptibility to, somatic}, 137800
IDH2	99,7	97,4	100	99,8	D-2-hydroxyglutaric aciduria 2, 613657
IDS	99,9	98	100	100	Mucopolysaccharidosis II, 309900

IDUA	93,7	86,8	100	100	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IFIH1	99,7	98,4	100	100	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	99,3	95,6	100	100	Osteogenesis imperfecta, type V, 610967
IFT122	100	99,6	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	100	100	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	100	99,9	100	100	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	97,6	88,2	100	100	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	93,5	90,1	95	94,9	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100	99,9	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100	99,9	100	100	Insulin-like growth factor I, resistance to, 270450
IGF2	100	100	100	100	Silver-Russell syndrome 3, 616489
IGFALS	99,9	99,6	100	100	Acid-labile subunit, deficiency of, 615961
IGSF1	99,5	96,3	100	100	Hypothyroidism, central, and testicular enlargement, 300888
IHH	100	100	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	99,8	97,4	100	100	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	84,1	77,2	100	100	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291
IL1RN	100	100	100	100	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852
IL2RG	99,8	97,1	100	100	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL6ST	96,4	90,3	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IMPAD1	100	100	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INPPL1	98,4	94,5	99,9	99,7	Opsismodysplasia, 258480

INTU	99,7	98,1	100	100	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
KAT6B	99,6	98,3	100	100	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNJ2	100	100	100	100	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KDELR2	100	100	100	100	Osteogenesis imperfecta 21, 619131
KIAA0586	97,3	93,1	95,8	95,8	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	100	99,3	100	100	?Orofaciodigital syndrome XV, 617127
KIF22	100	100	100	100	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KL	98,2	97,2	98,5	97,5	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KMT2A	100	99,9	99,9	99,4	Wiedemann-Steiner syndrome, 605130
KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 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	99,4	94,5	100	100	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LBX1	100	100	100	100	No OMIM disease ID
LEMD3	99,9	98,7	100	100	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
LFNG	87,9	86,4	92,2	87,7	Spondylocostal dysostosis 3, autosomal recessive, 609813

LHX3	96,6	96,5	100	100	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100	100	100	100	Pituitary hormone deficiency, combined, 4, 262700
LIFR	99,7	98	100	100	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMNA	97,4	91,9	100	100	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	99,6	96,3	100	100	Nail-patella syndrome, 161200 Focal segmental glomerulosclerosis 10, 256020
LONP1	100	99,8	100	100	CODAS syndrome, 600373
LPIN2	100	100	100	100	Majeed syndrome, 609628
LRP4	99,1	98,8	100	100	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	98,5	98,1	100	99,7	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 {Osteoporosis}, 166710 [Bone mineral density variability 1], 601884
LRRK1	98,6	97,5	100	100	No OMIM disease ID
LTBP2	99,9	99	100	100	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	99,6	98,1	100	100	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809

LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAFB	100	99,4	100	100	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAN2B1	99,8	97,9	100	100	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,8	86,5	100	100	Mannosidosis, beta, 248510
MAP2K1	99,8	97,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,1	100	100	Cardiofaciocutaneous syndrome 4, 615280
MAP3K20	100	99,5	100	100	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAP3K7	100	99,6	100	100	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MATN3	84,7	84,6	100	100	{Osteoarthritis susceptibility 2}, 140600 ?Spondyloepimetaphyseal dysplasia, Borochowitz Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBTPS2	100	99	100	100	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918
MECOM	100	99,9	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MEGF8	99,9	99	100	100	Carpenter syndrome 2, 614976
MEOX1	100	98,9	100	100	Klippel-Feil syndrome 2, 214300
MESD	100	99,9	100	100	Osteogenesis imperfecta, type XX, 618644
MESP2	93,9	86,9	97,5	97,5	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100	99,5	100	100	{Osteofibrous dysplasia, susceptibility to}, 607278 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MGP	98,7	95,1	100	100	Keutel syndrome, 245150
MIR140	NC	NC	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MMP13	95,2	92,2	92,4	92,4	Metaphyseal dysplasia, Spahr type, 250400 Metaphyseal anadysplasia 1, 602111 ?Spondyloepimetaphyseal dysplasia, Missouri type, 602111

MMP14	100	98,9	100	100	?Winchester syndrome, 277950
MMP2	100	100	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	99,1	96,1	100	100	Metaphyseal anadysplasia 2, 613073
MNX1	68,2	58,3	87,4	79,2	Currarino syndrome, 176450
MSX2	100	99,4	100	100	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	99,1	93,5	100	100	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYCN	100	99,9	99,3	96,7	Feingold syndrome 1, 164280
MYH3	99,9	99	100	100	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110
MYLPF	100	100	100	100	Arthrogryposis, distal, type 1C, 619110
MYO18B	100	99,1	100	100	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
NAGLU	92,9	89,9	99,9	99,2	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	100	99,6	100	100	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEK1	99,8	98	100	100	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	100	99,6	100	100	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	99,7	97,7	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NF1	92,6	90,2	100	100	Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Watson syndrome, 193520 Neurofibromatosis, type 1, 162200
NFIX	100	99,5	99,6	98,7	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NIN	100	99,5	99,1	99,1	?Seckel syndrome 7, 614851
NIPBL	98,9	97	100	100	Cornelia de Lange syndrome 1, 122470
NKX3-2	99,8	97	100	100	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330

NLRP3	100	99,9	100	100	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	100	100	100	100	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	99,2	97,2	100	100	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	100	99,5	100	100	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPPC	100	99	100	100	No OMIM disease ID
NPR2	100	99,6	100	100	Short stature with nonspecific skeletal abnormalities, 616255 Epiphyseal chondrodysplasia, Miura type, 615923 Acromesomelic dysplasia, Maroteaux type, 602875
NPR3	100	100	100	100	?Hypertension, salt-resistant, 0
NRAS	100	100	100	100	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	100	99,9	100	100	Sotos syndrome 1, 117550
NSDHL	100	98,7	100	100	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	99,7	98,2	100	100	Seckel syndrome 10, 617253
NXN	100	100	99,9	99,5	Robinow syndrome, autosomal recessive 2, 618529
OBSL1	100	99,3	100	100	3-M syndrome 2, 612921
OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
ORC1	100	99,4	100	100	Meier-Gorlin syndrome 1, 224690

ORC4	98,7	93,6	100	100	Meier-Gorlin syndrome 2, 613800
ORC6	100	99,9	100	100	Meier-Gorlin syndrome 3, 613803
OSTM1	98,6	94	100	100	Osteopetrosis, autosomal recessive 5, 259720
OTX2	100	99,7	100	100	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
P3H1	100	100	100	100	Osteogenesis imperfecta, type VIII, 610915
P4HB	94,6	94	100	100	Cole-Carpenter syndrome 1, 112240
PAM16	65,3	65,2	82,9	82,9	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	100	99,9	100	100	No OMIM disease ID
PAPSS2	100	99,5	100	100	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PAX3	100	99,9	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCNT	99,6	97,1	100	100	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	98,9	95,5	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE3A	99,9	99,4	100	100	Hypertension and brachydactyly syndrome, 112410
PDE4D	95,7	93,5	100	99,8	Acrodysostosis 2, with or without hormone resistance, 614613
PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	100	99,6	99,9	99,2	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3R1	99,8	99	100	100	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PISD	100	100	100	100	Liberfarb syndrome, 618889

PITX1	96,7	92	100	100	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	99,9	97,7	100	100	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PKDCC	90,6	81,5	97,8	94,7	Rhizomelic limb shortening with dysmorphic features, 618821
PLAG1	100	100	100	100	Silver-Russell syndrome 4, 618907 Adenomas, salivary gland pleomorphic, somatic, 181030
PLCB3	100	99	100	100	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	99,9	98,8	100	100	Auriculocondylar syndrome 2, 614669
PLEKHM1	100	99,8	100	100	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLK4	99,9	98,2	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100	98,4	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,3	97,3	100	100	Bruck syndrome 2, 609220
PLS3	97,7	96,1	97,2	97,2	Bone mineral density QTL18, osteoporosis, 300910
PNPLA6	100	99,7	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POC1A	100	100	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POLE	100	99,8	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336 {Colorectal cancer, susceptibility to, 12}, 615083
POLL	100	99,2	100	100	No OMIM disease ID
POLR1A	100	99,4	100	100	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	90,5	87	82,8	82,8	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,6	91,6	100	100	Treacher Collins syndrome 2, 613717
POLR3A	100	99,7	100	100	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,9	98,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POP1	100	99,7	100	100	Anauxetic dysplasia 2, 617396

POR	99,8	98,6	100	100	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
POU1F1	100	99,2	100	100	Pituitary hormone deficiency, combined, 1, 613038
PPIB	100	100	100	100	Osteogenesis imperfecta, type IX, 259440
PPM1D	100	99,9	100	100	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPP1CB	99,9	99,3	100	100	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PRKAR1A	99,3	93,5	100	100	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
PRKG2	98,2	96,7	100	100	No OMIM disease ID
PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	92,6	82,6	100	100	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	95,3	81,6	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMB1	100	100	100	100	No OMIM disease ID
PTDSS1	100	100	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	100	98,7	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTHLH	99,7	98,4	100	100	Brachydactyly, type E2, 613382
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PYCR1	99,9	97,7	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	100	99,5	100	100	Carpenter syndrome, 201000
RAB33B	85	85	100	100	Smith-McCort dysplasia 2, 615222
RAC3	97,3	94,4	99,7	98,2	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	99,2	96,6	100	100	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAF1	100	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916

RASGRP2	99,7	97,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RBBP8	100	99,7	100	100	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBM8A	99,8	97,9	100	100	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	98,4	92,8	100	100	Adams-Oliver syndrome 3, 614814
RECQL4	99,8	98,1	100	99,9	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400
RIPPLY2	100	97,9	100	100	?Spondylocostal dysostosis 6, 616566
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNPC3	91,5	70,7	100	100	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	NC	Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROR2	100	99,9	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPL10	97,4	89,1	100	100	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPL13	96,3	85,5	100	100	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RRAS	99,8	95,7	100	100	No OMIM disease ID
RREB1	99,9	99,2	100	100	No OMIM disease ID
RSPO2	97,1	90,7	100	100	Tetraamelia syndrome 2, 618021 ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022
RSPRY1	100	100	100	100	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	72,2	72,2	100	100	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	99,9	99	100	100	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480

SALL4	98,6	96,7	100	100	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SBDS	100	100	100	100	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400
SCARF2	95,4	86,2	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SEC24D	100	99,7	100	100	Cole-Carpenter syndrome 2, 616294
SEMA3A	100	99,9	100	100	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SERPINF1	100	100	100	100	Osteogenesis imperfecta, type VI, 613982
SERPINH1	100	98,3	100	100	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SETD2	100	99,9	100	100	Luscan-Lumish syndrome, 616831
SF3B4	99,9	97,3	100	100	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100	99,8	100	100	Pyle disease, 265900
SGMS2	100	100	100	100	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGSH	94,4	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3BP2	91,4	91,2	97	95,3	Cherubism, 118400
SH3PXD2B	100	100	100	100	Frank-ter Haar syndrome, 249420
SHH	100	99,5	100	100	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	99,9	99,4	100	100	Noonan syndrome-like with loose anagen hair 1, 607721
SHOX	70	59,7	95,1	95,1	Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300
SKI	99,3	94,9	100	99,4	Shprintzen-Goldberg syndrome, 182212
SLC10A7	99,7	98	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC17A5	99,6	97	100	100	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC25A24	99,4	99,3	99,8	99,8	Fontaine progeroid syndrome, 612289
SLC26A2	100	100	100	100	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	100	99,6	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782

SLC34A3	100	99,4	100	100	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35C1	99,9	98,7	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100	97,7	100	100	Schneckenbecken dysplasia, 269250
SLC39A13	99,8	98,2	100	100	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLCO2A1	100	99,4	100	100	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	99,7	98,8	100	100	No OMIM disease ID
SMAD2	100	99,6	100	100	No OMIM disease ID
SMAD3	99,9	99	100	100	Loeys-Dietz syndrome 3, 613795
SMAD4	100	99,9	100	100	Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350
SMARCA4	99,9	99	100	100	{Rhabdoid tumor predisposition syndrome 2}, 613325 Coffin-Siris syndrome 4, 614609
SMARCAL1	100	99,9	100	100	Schimke immunoosseous dysplasia, 242900
SMARCB1	100	100	100	100	Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091 Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCE1	95,6	88,8	100	100	{Meningioma, familial, susceptibility to}, 607174 Coffin-Siris syndrome 5, 616938
SMC1A	100	98,7	100	99,8	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	95,2	91	100	100	Cornelia de Lange syndrome 3, 610759
SNRPB	100	99,3	100	100	Cerebrocostomandibular syndrome, 117650
SNX10	96,2	95,7	100	99,6	Osteopetrosis, autosomal recessive 8, 615085
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100	99,2	100	100	Noonan syndrome 9, 616559
SOST	100	99,5	100	100	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX2	100	100	100	100	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	91,4	75,2	100	99,5	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123

SOX9	100	98,6	100	100	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP7	100	99,8	100	100	Osteogenesis imperfecta, type XII, 613849
SPARC	100	100	100	100	Osteogenesis imperfecta, type XVII, 616507
SPECC1L	96	95,7	97,8	96,2	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPINK5	99,9	99,5	100	100	Netherton syndrome, 256500
SPR	99,8	96,3	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100	98,9	100	100	Legius syndrome, 611431
SRCAP	99,4	98,9	100	100	Floating-Harbor syndrome, 136140
SRP54	99,5	96,5	100	100	Neutropenia, severe congenital, 8, autosomal dominant, 618752
STAT3	100	99,8	100	100	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	100	98,5	100	100	Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Leukemia, acute promyelocytic, somatic, 102578
SULF1	99,9	99,3	100	100	No OMIM disease ID
SUMF1	97,5	90,8	100	100	Multiple sulfatase deficiency, 272200
TAB2	100	99,7	100	100	Congenital heart defects, nonsyndromic, 2, 614980
TAPT1	91,7	86,9	98,5	94,8	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBCE	99,8	97,5	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBX15	100	99,9	100	100	Cousin syndrome, 260660
TBX3	99,2	96,8	100	100	Ulnar-mammary syndrome, 181450
TBX4	97,6	95,1	100	100	Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891
TBX5	100	100	100	100	Holt-Oram syndrome, 142900
TBX6	99,5	95,5	100	100	Spondylocostal dysostosis 5, 122600
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TCF12	100	99,9	100	100	Craniosynostosis 3, 615314
TCIRG1	97,6	90,1	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCOF1	99,7	98,6	100	100	Treacher Collins syndrome 1, 154500
TCTEX1D2	100	100	100	100	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405

TCTN2	100	99,5	100	100	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100	100	100	100	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TENT5A	100	99,7	100	100	Osteogenesis imperfecta, type XVIII, 617952
TGDS	99,4	96,8	100	100	Catel-Manzke syndrome, 616145
TGFB1	100	99,9	100	100	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
TGFB2	100	100	100	100	Loeys-Dietz syndrome 4, 614816
TGFB3	100	100	100	100	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBR1	93,7	93,6	99	96,3	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	100	100	100	100	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
THPO	81,4	81	100	100	Thrombocythemia 1, 187950
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100	99,6	100	100	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM251	100	99,5	100	100	No OMIM disease ID
TMEM38B	100	99,9	100	100	Osteogenesis imperfecta, type XIV, 615066
TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TNFRSF11A	94,6	93,3	99,2	98	Osteolysis, familial expansile, 174810 {Paget disease of bone 2, early-onset}, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	100	100	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	100	99,9	100	100	Osteopetrosis, autosomal recessive 2, 259710
TONSL	99,8	97,8	100	100	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510

TP63	100	100	100	100	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	99,6	97,6	100	100	Senior-Loken syndrome 9, 616629
TRAIP	100	100	100	100	Seckel syndrome 9, 616777
TRAPPC2	89,7	69,6	100	100	Spondyloepiphyseal dysplasia tarda, 313400
TREM2	100	99,8	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TRIP11	98,4	94	100	100	Osteochondrodysplasia, 184260 Achondrogenesis, type IA, 200600
TRPS1	100	99,9	100	100	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV4	100	99,9	100	100	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 [Sodium serum level QTL 1], 613508 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	100	99,5	99,9	98,9	Hyperparathyroidism, transient neonatal, 618188
TTC21B	99,9	99,3	100	100	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTI2	100	100	100	100	Mental retardation, autosomal recessive 39, 615541
TWIST1	100	98,9	97,2	92,3	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TYROBP	100	100	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
UFSP2	100	99,6	100	100	?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 ?Hip dysplasia, Beukes type, 142669
VAC14	99,9	98,5	100	100	Striatonigral degeneration, childhood-onset, 617054

VDR	97,2	94,9	98,2	95,2	Rickets, vitamin D-resistant, type IIA, 277440
VPS33A	91,3	89,8	89,9	89,9	Mucopolysaccharidosis-plus syndrome, 617303
VPS35L	100	99,9	100	100	Ritscher-Schinzel syndrome 3, 619135
WDR19	100	99,4	100	100	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	100	99,6	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	99,8	98,9	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	99,5	97	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNT1	99,3	95,3	100	100	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 Osteogenesis imperfecta, type XV, 615220
WNT10B	100	99,4	100	100	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT3	100	99,6	100	100	?Tetra-amelia syndrome 1, 273395
WNT5A	100	100	100	100	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100	98,7	100	100	No OMIM disease ID
WNT7A	100	100	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
XRCC4	99,9	99,3	100	100	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,4	89,6	98,1	94,8	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Desbuquois dysplasia 2, 615777
XYLT2	100	98,3	96,7	96,7	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822
ZBTB16	100	99,9	100	100	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZMPSTE24	100	99,9	100	100	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZSWIM6	95,5	91,9	94,9	92,1	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-protein coding genes.
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 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors