SHORT STATURE AND SKELETAL DYSPLASIA GENE PANEL DG 3.2.0 (557 genes)

Releasedate: 16-09-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype Description and OMIM disease ID
ABCC9	100	99,8	100	100	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050
ACAN	96,9	92,5	98,9	98,7	?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	99,9	98,9	100	100	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	99,9	97,2	100	100	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACVR1	100	99,9	100	100	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	100	99,9	100	100	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	92,9	88,8	97,9	95,9	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	98	94,7	99,9	99,8	Geleophysic dysplasia 1, 231050
AFF3	98,6	97,9	100	100	KINSSHIP syndrome, 619297
AGA	100	99,9	100	100	Aspartylglucosaminuria, 208400
AGPS	98,8	95,2	100	99,4	Rhizomelic chondrodysplasia punctata, type 3, 600121
AIFM1	99,9	97,8	100	100	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
ALG12	100	99,9	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG3	100	99,5	100	100	Congenital disorder of glycosylation, type Id, 601110

ALG9	99,9	99,3	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	99,7	99,5	100	100	Alstrom syndrome, 203800
ALPL	100	99,4	100	100	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
ALX1	99,6	95,2	100	100	Frontonasal dysplasia 3, 613456
ALX3	80,2	72,8	100	100	Frontonasal dysplasia 1, 136760
ALX4	100	99,9	100	100	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMER1	99,6	96,6	100	100	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	99,9	98,4	100	100	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKH	100	99,9	100	100	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	97	94	100	100	KBG syndrome, 148050
ANO5	99,2	96,9	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260
ANTXR2	99,5	97,2	100	100	Hyaline fibromatosis syndrome, 228600
APC2	98,3	94,8	99,7	98,5	Cortical dysplasia, complex, with other brain malformations 10, 618677 ?Sotos syndrome 3, 617169
ARCN1	96,8	96,6	96,7	96,6	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARHGAP31	99,7	98,2	100	100	Adams-Oliver syndrome 1, 100300
ARID1B	96,2	94,6	97,8	96,9	Coffin-Siris syndrome 1, 135900
ARSB	98,8	91	100	100	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSL	98,9	92,4	100	99,8	Chondrodysplasia punctata, X-linked recessive, 302950
ATP6V0A2	99,9	98,7	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATR	99,7	99	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
B3GALT6	77	73	91,7	81	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465

B3GAT3	99,4	96,6	95,4	94,8	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	99,7	96,8	100	99,4	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BGN	100	99,9	100	100	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	72,5	53,5	99,6	96,9	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BMP1	100	100	100	100	Osteogenesis imperfecta, type XIII, 614856
BMP2	100	100	100	100	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BMPER	100	99,6	100	100	Diaphanospondylodysostosis, 608022
BMPR1B	99,9	99,9	100	100	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849
IMPAD1	100	99,9	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BRAF	89,4	77,6	100	100	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	99,8	98,4	100	100	Cerebellofaciodental syndrome, 616202
BTK	100	99,7	100	99,8	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	97,6	97,2	100	100	No OMIM disease ID
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CANT1	100	100	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CASR	100	99,5	100	100	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980
CBL	97,3	96,9	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785

CC2D2A	98,3	96,6	97,1	97	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCDC134	100	100	100	100	No OMIM disease ID
CCDC8	100	100	100	100	3-M syndrome 3, 614205
CCN6	84,6	84,6	84,9	84,6	Progressive pseudorheumatoid dysplasia, 208230
CCNQ	82,9	78,3	99,8	98,2	STAR syndrome, 300707
CDC42	96,3	87,9	100	100	Takenouchi-Kosaki syndrome, 616737
CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063
CDC6	100	99,9	100	100	?Meier-Gorlin syndrome 5, 613805
CDC73	99,8	98,3	100	100	Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism-jaw tumor syndrome, 145001
CDKN1C	89,9	81,6	98,9	95,8	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDT1	99,7	97,8	99,9	98	Meier-Gorlin syndrome 4, 613804
CENPE	96,7	90	100	99,7	?Microcephaly 13, primary, autosomal recessive, 616051
CEP120	99,9	99,6	100	100	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP152	99,5	98	100	100	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	96,2	90,8	100	99,9	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CFAP410	100	99,6	100	100	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
CHST11	100	100	100	100	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	99,9	98,8	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100	99,9	100	100	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHSY1	97,8	96,3	99,3	96,9	Temtamy preaxial brachydactyly syndrome, 605282
CILK1	99,6	98	100	99,8	Endocrine-cerebroosteodysplasia, 612651

CKAP2L	99,5	98,3	100	100	Filippi syndrome, 272440
CLCN5	99,7	97,1	100	99,9	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Hypophosphatemic rickets, 300554 Dent disease 1, 300009 Nephrolithiasis, type I, 310468
CLCN7	99,4	97,8	100	100	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
COG1	100	99,9	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG4	100	100	100	100	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COL10A1	100	99,6	100	100	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	96	92,7	100	99,9	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	100	99,6	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	99,8	98,2	100	100	Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL1A2	98,7	95,7	100	100	Osteogenesis imperfecta, type III, 259420 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	99,8	99,4	100	100	Steel syndrome, 615155
COL2A1	100	99,8	100	100	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 Czech dysplasia, 609162

					Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 SMED Strudwick type, 184250 Stickler sydrome, type I, nonsyndromic ocular, 609508 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Kniest dysplasia, 156550 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	99,9	98,6	100	100	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	99,9	98,9	100	100	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COL9A3	98,9	95,5	99,9	99,3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COMP	93,8	92,4	100	100	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400
CPLANE1	99,4	98,2	100	100	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CREB3L1	100	99,9	100	100	Osteogenesis imperfecta, type XVI, 616229
CREBBP	99,6	97,8	100	100	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRIPT	98,9	93,5	100	100	Short stature with microcephaly and distinctive facies, 615789
CRTAP	100	99,4	100	100	Osteogenesis imperfecta, type VII, 610682
CSF1R	100	99,6	100	100	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSGALNACT1	100	100	100	100	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CTSA	100	99,6	100	100	Galactosialidosis, 256540
CTSK	100	99,2	100	100	Pycnodysostosis, 265800
CUL7	100	99,1	100	100	3-M syndrome 1, 273750

CYP26B1	100	100	100	100	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	100	99,8	100	100	Vitamin D-dependent rickets, type I, 264700
CYP2R1	99,5	96	100	100	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
DDR2	100	99,6	100	100	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100	100	100	100	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX58	99,6	98,3	100	99,9	Singleton-Merten syndrome 2, 616298
DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHODH	100	99,9	100	100	Miller syndrome, 263750
DLL3	93	87,8	100	99,5	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100	99,4	100	100	Adams-Oliver syndrome 6, 616589
DLX3	99,8	97,6	100	100	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX5	99,9	98,2	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	99,9	99,9	100	100	Hypophosphatemic rickets, AR, 241520
DNA2	99,6	96,9	100	100	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJC21	99,5	97,4	100	100	Bone marrow failure syndrome 3, 617052
DNMT3A	99,8	98,2	100	100	Tatton-Brown-Rahman syndrome, 615879 Acute myeloid leukemia, somatic, 601626 Heyn-Sproul-Jackson syndrome, 618724
DOCK6	99,4	98,7	100	100	Adams-Oliver syndrome 2, 614219
DONSON	93,8	85,8	100	100	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPCD	100	100	100	100	No OMIM disease ID
DPM1	97,4	90,9	98,6	94,6	Congenital disorder of glycosylation, type Ie, 608799
DSE	98,4	95,4	100	100	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DVL1	97,2	95,1	100	100	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100	100	100	100	Robinow syndrome, autosomal dominant 3, 616894

DYM	97	95,6	100	100	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC2H1	98,6	95,2	100	99,8	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	99,3	95,8	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	100	99,8	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	99,6	98,4	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	100	99,8	100	100	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EBP	99,5	94,3	100	100	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECEL1	95,9	91,8	100	100	Arthrogryposis, distal, type 5D, 615065
EDN1	100	99,4	100	100	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706
EDNRA	99,8	99,8	100	99,9	Mandibulofacial dysostosis with alopecia, 616367
EFL1	99,3	97,7	100	100	Shwachman-Diamond syndrome 2, 617941
EFNB1	100	100	100	100	Craniofrontonasal dysplasia, 304110
EFTUD2	100	99,2	100	100	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2AK3	98,2	95,5	100	99,8	Wolcott-Rallison syndrome, 226980
EIF4A3	100	99,2	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
ENPP1	96,5	90,6	98,8	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
EOGT	79,3	77,8	91,8	88,3	Adams-Oliver syndrome 4, 615297
EP300	99,9	98,9	100	100	Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
ERF	100	98,9	100	100	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ESCO2	98,5	94,6	100	99,7	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
EVC	94,2	91,4	97,5	95,1	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	98	96,2	100	100	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530

EXOC6B	98,1	97,1	99,9	99,5	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC5	100	100	100	100	No OMIM disease ID
EXT1	99,6	97,1	100	100	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	99,9	99	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	99,7	98	100	100	Weaver syndrome, 277590
FAM111A	100	99,4	100	100	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361
FAM20B	100	99,4	100	100	No OMIM disease ID
FAM20C	100	100	100	99,7	Raine syndrome, 259775
FAR1	97,4	94	100	100	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FBLN1	99,7	96,6	100	99,9	No OMIM disease ID
FBN1	100	99,7	100	100	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	100	99,8	100	100	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FBXW4	82,2	79,8	88,8	84,2	No OMIM disease ID
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FGD1	97,2	91,2	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	100	99,5	100	100	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	99,4	96,7	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF8	97,1	87,2	100	99,9	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

FGF9	100	100	100	100	Multiple synostoses syndrome 3, 612961
FGFR1	100	99,3	100	100	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	97,6	97	100	100	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Jackson-Weiss syndrome, 123150 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Apert syndrome, 101200 Pfeiffer syndrome, 101600 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,
FGFR3	99,8	98	100	100	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 LADD syndrome, 149730 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 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247
FIG4	99,8	99,7	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691

					Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FKBP10	98,9	97,3	100	100	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450
FKBP14	99,8	98,7	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLNA	100	99,9	100	100	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	99,4	98,7	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FMN1	97,1	95,7	100	100	No OMIM disease ID
FN1	99,9	98,7	100	100	Spondylometaphyseal dysplasia, corner fracture type, 184255 Glomerulopathy with fibronectin deposits 2, 601894
FUCA1	100	100	100	100	Fucosidosis, 230000
FUZ	100	100	100	100	No OMIM disease ID
FZD2	100	97,8	100	100	Omodysplasia 2, 164745
GALNS	100	99,3	100	100	Mucopolysaccharidosis IVA, 253000
GALNT3	99,8	98,7	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GCM2	100	100	100	100	Hypoparathyroidism, familial isolated 2, 618883 Hyperparathyroidism 4, 617343
GDF3	100	100	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	100	100	100	100	Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298

					?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A1, C, 615072
GDF6	100	100	100	99,6	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	100	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	99,5	99,5	99,5	99,5	Laron dwarfism, 262500 Increased responsiveness to growth hormone, 604271 Growth hormone insensitivity, partial, 604271
GHRHR	96,5	96,4	100	99,9	Growth hormone deficiency, isolated, type IV, 618157
GHSR	98,7	95,6	100	100	Growth hormone deficiency, isolated partial, 615925
GINS2	100	97,5	100	100	No OMIM disease ID
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GLB1	99,2	92,8	100	100	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLI2	99,8	98,6	100	99,9	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	98,5	97,7	100	100	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200

					Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GMNN	99,6	96,3	100	99,8	Meier-Gorlin syndrome 6, 616835
GNAI3	98,4	93,2	100	100	Auriculocondylar syndrome 1, 602483
GNAS	86,7	84,4	81,8	81,7	ACTH-independent macronodular adrenal hyperplasia, 219080 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ia, 103580 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 McCune-Albright syndrome, somatic, mosaic, 174800 Pseudopseudohypoparathyroidism, 612463
GNPAT	99,5	95,6	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	69,3	46,4	100	100	No OMIM disease ID
GNPTAB	99,9	99,7	100	100	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	99,8	96,6	100	100	Mucolipidosis III gamma, 252605
GNS	99,2	94,6	100	100	Mucopolysaccharidosis type IIID, 252940
GORAB	99,7	97,2	100	100	Geroderma osteodysplasticum, 231070
GPC3	98,8	92,9	100	99,9	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC6	100	99,9	100	100	Omodysplasia 1, 258315
GPR161	100	100	100	100	No OMIM disease ID
GPX4	91,3	88,4	98,8	96,1	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GSC	98,9	93	100	100	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GUSB	92,5	90,1	100	100	Mucopolysaccharidosis VII, 253220
GZF1	100	99,7	100	100	Joint laxity, short stature, and myopia, 617662
H19	NC	NC	NC	NC	No OMIM disease ID
HAAO	100	100	100	100	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HDAC4	100	99,9	100	100	No OMIM disease ID
HDAC8	85,7	83,7	96,4	95,2	Cornelia de Lange syndrome 5, 300882
HES7	75,6	44,1	100	100	Spondylocostal dysostosis 4, autosomal recessive, 613686

HESX1	99,3	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	86,4	86,2	91,3	89,1	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HMGA2	81	76,6	89,6	80,1	Silver-Russell syndrome 5, 618908
HOXA11	97,1	88,3	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	76,4	67,3	90,3	81,4	Hand-foot-uterus syndrome, 140000 ?Guttmacher syndrome, 176305
HOXD13	100	98,8	100	100	Syndactyly, type V, 186300 Synpolydactyly 1, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 ?Brachydactyly-syndactyly syndrome, 610713
HPGD	99,5	99,3	100	99,7	?Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HRAS	100	100	100	100	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
HSPA9	87,1	82,8	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPG2	99,2	97,5	100	99,8	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IARS2	99,9	99,8	100	100	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
ID4	90,7	85,3	98,8	90,6	No OMIM disease ID
IDH1	90,6	75,5	100	100	No OMIM disease ID
IDH2	99,8	97,4	100	100	D-2-hydroxyglutaric aciduria 2, 613657
IDS	99,6	95,3	100	100	Mucopolysaccharidosis II, 309900

IDUA	94,6	87,4	100	100	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IFIH1	99,5	97,3	100	100	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	99,7	97	100	100	Osteogenesis imperfecta, type V, 610967
IFT122	99,9	99,2	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,9	99,2	100	100	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	99,6	98,6	100	100	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100	99,9	100	99,9	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	97,2	85,7	100	99,9	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92,9	89,6	94,9	94,6	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	99,8	99,8	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100	99,7	100	100	Insulin-like growth factor I, resistance to, 270450
IGF2	100	100	100	100	Silver-Russell syndrome 3, 616489
IGFALS	100	100	100	100	Acid-labile subunit, deficiency of, 615961
IGSF1	99,3	94	100	100	Hypothyroidism, central, and testicular enlargement, 300888
IHH	100	100	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	99	96,1	100	100	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IKBKG	84,6	75,2	100	100	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IL1RN	100	99,9	100	100	Interleukin 1 receptor antagonist deficiency, 612852
IL2RG	99,8	92,8	100	100	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL6ST	94,9	89,4	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523

INPPL1	98,6	94,4	100	99,9	Opsismodysplasia, 258480
INTU	99,9	98,6	100	100	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
KAT6B	99,4	98	100	100	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNJ2	100	100	100	100	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KDELR2	100	100	100	100	Osteogenesis imperfecta 21, 619131
KIAA0586	97,1	92	95,8	95,7	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA0753	99,9	98,9	100	100	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIF22	100	99,6	100	100	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	93,6	91,9	99,7	98,6	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KL	98,5	97,5	98,7	97,9	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KMT2A	100	99,7	100	99,8	Wiedemann-Steiner syndrome, 605130
KRAS	99	97,8	100	100	Gastric cancer, somatic, 137215 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	99,5	96,7	100	100	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
LAMA5	98,5	96,3	100	99,9	No OMIM disease ID

LBR	97,9	91	100	100	Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019 ?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140
LBX1	100	100	100	100	No OMIM disease ID
LEMD3	99,5	97,8	100	100	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700
LFNG	88,6	86,5	92	87,3	Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	96,6	96,2	100	100	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100	100	100	100	Pituitary hormone deficiency, combined, 4, 262700
LIFR	99,3	97,8	100	99,9	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMNA	96,1	90,6	100	100	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Restrictive dermopathy, lethal, 275210 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 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	99,3	96,8	100	100	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LONP1	100	99,9	100	100	CODAS syndrome, 600373
LPIN2	99,9	99,7	100	100	Majeed syndrome, 609628
LRP4	99,1	98,4	100	100	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	99,2	98,2	99,8	99,2	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636

LRRK1	98,7	97,5	100	100	No OMIM disease ID
LTBP1	99,2	97,4	100	99,9	Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	99,8	98,9	100	100	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	99,8	98,6	100	99,9	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	100	99,9	100	100	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAFB	100	99,8	100	100	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAN2B1	99,6	97,4	100	100	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,1	84,9	100	99,9	Mannosidosis, beta, 248510
MAP2K1	99,6	96,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,3	100	100	Cardiofaciocutaneous syndrome 4, 615280
MAP3K20	99,9	99,2	100	99,9	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	99,8	99,6	100	100	Frontometaphyseal dysplasia 2, 617137 Cardiospondylocarpofacial syndrome, 157800
MAPK1	100	99,9	100	99,6	Noonan syndrome 13, 619087
MATN3	84,8	84,1	100	100	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBTPS2	99,9	98,5	100	100	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	99,7	100	100	?Meier-Gorlin syndrome 8, 617564
MECOM	100	99,6	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MEGF8	100	99,2	100	100	Carpenter syndrome 2, 614976
MEOX1	100	98	100	100	Klippel-Feil syndrome 2, 214300
MESD	100	98,8	100	100	Osteogenesis imperfecta, type XX, 618644
MESP2	95,7	90,6	97,5	97,5	Spondylocostal dysostosis 2, autosomal recessive, 608681

MET	100	99,4	100	100	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705
MGP	98,7	93,6	100	100	Keutel syndrome, 245150
MIR140	NC	NC	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MKS1	99,4	96,3	100	100	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MMP13	93,6	92,2	92,4	92,3	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400
MMP14	100	99,4	100	100	?Winchester syndrome, 277950
MMP2	100	100	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	99,6	96,8	100	100	Metaphyseal anadysplasia 2, 613073
MNX1	70,8	59,2	85,7	79	Currarino syndrome, 176450
MRAS	100	99,3	100	100	Noonan syndrome 11, 618499
MSX2	100	98	100	100	Parietal foramina with cleidocranial dysplasia, 168550 Craniosynostosis 2, 604757 Parietal foramina 1, 168500
MTAP	98,3	91,8	100	100	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYCN	100	100	99,2	96,2	Feingold syndrome 1, 164280
MYH3	99,9	98,4	100	100	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
MYLPF	100	100	100	100	Arthrogryposis, distal, type 1C, 619110
MYO18B	100	99,3	100	100	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
NADSYN1	100	100	100	100	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGLU	93,8	91,7	99,9	98,7	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	99,9	99,3	100	100	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NEK1	99,5	98,2	100	99,9	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520

NEK9	99,9	99	100	100	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022
NEU1	99,3	96,1	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NF1	91,8	89,3	100	100	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFIX	100	99,3	99,4	98,6	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NIN	99,9	99,5	99,1	99,1	?Seckel syndrome 7, 614851
NIPBL	98,4	96,3	100	99,9	Cornelia de Lange syndrome 1, 122470
NKX3-2	100	99,3	100	100	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP3	100	99,9	100	100	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
NOG	100	100	100	100	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	99,3	97,9	100	100	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	100	99,2	100	100	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPC	100	99,3	100	100	No OMIM disease ID
NPR2	100	99,2	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NPR3	100	100	100	100	No OMIM disease ID
NRAS	100	100	100	100	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	99,8	100	100	Sotos syndrome 1, 117550
NSD2	99,9	98,3	100	100	No OMIM disease ID
NSDHL	99,8	96,3	100	100	CK syndrome, 300831 CHILD syndrome, 308050
NSMCE2	99,5	98,7	100	100	Seckel syndrome 10, 617253
NXN	100	100	100	99,7	Robinow syndrome, autosomal recessive 2, 618529
OBSL1	100	99,8	100	100	3-M syndrome 2, 612921
OFD1	87,1	71,3	100	99,8	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
ORC1	99,9	97,9	100	100	Meier-Gorlin syndrome 1, 224690
ORC4	96,8	90,6	100	100	Meier-Gorlin syndrome 2, 613800
ORC6	100	99,8	100	100	Meier-Gorlin syndrome 3, 613803
OSTM1	98,7	92,9	100	100	Osteopetrosis, autosomal recessive 5, 259720
OTX2	100	99	100	100	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 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	99,6	100	100	Short stature, Dauber-Argente type, 619489
PAPSS2	99,8	98	100	100	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PAX3	100	99,8	100	100	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220

PCNT	99,3	96,5	100	100	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	99,2	95,7	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE3A	99,8	99,1	100	100	Hypertension and brachydactyly syndrome, 112410
PDE4D	95,7	93,1	100	99,6	Acrodysostosis 2, with or without hormone resistance, 614613
PEX5	99,9	98,8	100	100	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	96,4	88	100	100	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	88	81	91,3	91,2	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHEX	99,9	98,9	100	99,2	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	99,9	98,2	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3R1	99,7	98,4	100	100	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PISD	100	99,7	100	100	Liberfarb syndrome, 618889
PITX1	96,4	91,5	100	100	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	99,8	97,2	100	100	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PKDCC	91,7	84,5	98	94,6	Rhizomelic limb shortening with dysmorphic features, 618821
PLAG1	100	100	100	100	Adenomas, salivary gland pleomorphic, somatic, 181030 Silver-Russell syndrome 4, 618907
PLCB3	100	99,3	100	100	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	99,8	98,7	100	100	Auriculocondylar syndrome 2, 614669
PLEKHM1	100	99,9	100	100	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLK4	99,4	98,4	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100	98,2	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400

PLOD2	99,2	98,1	100	100	Bruck syndrome 2, 609220
PLS3	97,3	96,1	97,2	97,1	Bone mineral density QTL18, osteoporosis, 300910
PNPLA6	100	99,8	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
POC1A	100	100	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POLE	100	99,5	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLL	99,8	97,4	100	100	No OMIM disease ID
POLR1A	99,9	98,8	100	100	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	89,6	84,8	82,8	82,8	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	91,6	91,6	100	99,8	Treacher Collins syndrome 2, 613717
POLR3A	99,9	99	100	100	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	99,7	97,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POP1	100	99,3	100	100	Anauxetic dysplasia 2, 617396
POR	99,5	98	100	100	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
POU1F1	99,9	98,2	100	100	Pituitary hormone deficiency, combined, 1, 613038
PPIB	100	99,9	100	100	Osteogenesis imperfecta, type IX, 259440
PPM1D	100	99,9	100	100	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPP1CB	99,8	98,7	100	100	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R21	99,3	95,5	100	100	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PRKAR1A	97	89,1	100	100	Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Adrenocortical tumor, somatic,
PRKG2	97,6	96,7	100	100	No OMIM disease ID

PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	91	80,2	100	100	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	92	75,1	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMB1	100	99,8	100	100	No OMIM disease ID
PTDSS1	100	100	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	99,6	95,9	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
PTHLH	99,8	98,3	100	100	Brachydactyly, type E2, 613382
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PYCR1	100	98,2	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	99,7	99,7	100	100	Carpenter syndrome, 201000
RAB33B	85	85	100	100	Smith-McCort dysplasia 2, 615222
RAC3	98	94,3	99,4	97,3	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	99,2	95,9	100	100	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RAF1	99,9	99,2	100	100	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RALA	89,1	82,1	100	100	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RASGRP2	100	98,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RBBP8	99,7	99,4	100	99,9	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,
RBM8A	99,6	95,3	100	100	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	97,4	89	100	100	Adams-Oliver syndrome 3, 614814

RECQL4	99,9	98,6	100	100	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
RIPPLY2	99	94,7	100	99,9	?Spondylocostal dysostosis 6, 616566
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNPC3	94	75,1	100	100	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROR2	100	99,4	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	96,5	95,3	100	99,4	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPL10	96,7	87,5	100	100	Intellectual developmental disorder, X-linked, syndromic, 35, 300998
RPL13	95,6	84,7	100	100	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RRAS	99,7	95,8	100	99,9	No OMIM disease ID
RRAS2	95,9	87,4	100	100	Noonan syndrome 12, 618624 Ovarian carcinoma,
RREB1	99,8	99,2	100	100	No OMIM disease ID
RSPO2	94,8	88,3	100	100	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPRY1	99,9	99,9	100	100	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	72,2	72,2	100	100	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	99,7	97,5	100	100	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	99,1	96,4	100	100	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SATB2	99,5	96,5	100	100	Glass syndrome, 612313

SBDS	100	99,9	100	100	Shwachman-Diamond syndrome, 260400
SCARF2	97,4	88,9	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SEC24D	99,9	99,3	100	100	Cole-Carpenter syndrome 2, 616294
SEMA3A	100	99,7	100	100	No OMIM disease ID
SERPINF1	100	99,9	100	100	Osteogenesis imperfecta, type VI, 613982
SERPINH1	99,8	98	100	100	Osteogenesis imperfecta, type X, 613848
SETD2	99,9	99,6	100	100	Luscan-Lumish syndrome, 616831
SF3B4	99,8	94,1	100	100	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	99,8	99,1	100	100	Pyle disease, 265900
SGMS2	100	100	100	100	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGSH	94,8	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3BP2	91,4	91,3	97,2	95,9	Cherubism, 118400
SH3PXD2B	100	99,9	100	100	Frank-ter Haar syndrome, 249420
SHH	100	100	100	100	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	99,8	99,6	100	99,9	Noonan syndrome-like with loose anagen hair 1, 607721
SHOX	71,5	60,7	95,1	95,1	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	99,7	97,1	100	99,7	Shprintzen-Goldberg syndrome, 182212
SLC10A7	99,5	98,1	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC17A5	99,6	96,2	100	100	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC25A24	99,3	98,8	99,7	99,7	Fontaine progeroid syndrome, 612289
SLC26A2	100	100	100	100	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	99,5	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	100	99,3	100	100	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35C1	100	99,4	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	99,6	97,6	100	99,2	Schneckenbecken dysplasia, 269250
SLC39A13	99,9	97,9	100	100	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLCO2A1	99,9	98	100	100	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	99,3	98,2	100	100	No OMIM disease ID
SMAD2	100	99,8	100	100	No OMIM disease ID
SMAD3	99,9	98,4	100	100	Loeys-Dietz syndrome 3, 613795
SMAD4	99,9	99,9	100	100	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA4	99,9	99,2	100	100	Coffin-Siris syndrome 4, 614609
SMARCAL1	100	99,8	100	100	Schimke immunoosseous dysplasia, 242900
SMARCB1	100	99,9	100	100	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCE1	93,7	85,9	100	100	Coffin-Siris syndrome 5, 616938
SMC1A	99,6	97,1	100	99,9	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	94,5	89	100	99,9	Cornelia de Lange syndrome 3, 610759
SMOC2	76,7	74,9	100	100	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNRPB	100	98,6	100	100	Cerebrocostomandibular syndrome, 117650
SNX10	96,2	95,9	99,9	99,3	Osteopetrosis, autosomal recessive 8, 615085
SOS1	99,6	97,9	100	99,9	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	99,6	98,7	100	99,9	Noonan syndrome 9, 616559
SOST	100	99,6	100	100	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860

SOX2	100	99,8	100	100	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SOX3	94,9	81,2	100	99,6	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	100	99,9	100	100	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SP7	99,9	99,2	100	100	Osteogenesis imperfecta, type XII, 613849
SPARC	100	100	100	100	Osteogenesis imperfecta, type XVII, 616507
SPECC1L	96	95	97,1	96,1	Opitz GBBB syndrome, type II, 145410 Teebi hypertelorism syndrome, 145420 ?Facial clefting, oblique, 1, 600251
SPINK5	99,8	99,6	100	99,9	Netherton syndrome, 256500
SPR	100	99,4	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	99,8	98,2	100	100	Legius syndrome, 611431
SRCAP	99,7	98,9	100	100	Floating-Harbor syndrome, 136140
SRP54	98	93,4	100	100	Neutropenia, severe congenital, 8, autosomal dominant, 618752
STAT3	99,9	99	100	100	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	99,9	98,1	100	100	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Leukemia, acute promyelocytic, somatic, 102578
STIM1	99,9	97,5	100	100	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
SULF1	99,9	99,2	100	100	No OMIM disease ID
SUMF1	98,3	92,5	100	100	Multiple sulfatase deficiency, 272200
TAB2	99,8	99,2	100	100	Congenital heart defects, nonsyndromic, 2, 614980
TAPT1	93	87,1	98,5	94,1	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBCE	99,7	96,6	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBX15	100	99,7	100	100	Cousin syndrome, 260660
TBX3	99,4	97,3	100	100	Ulnar-mammary syndrome, 181450

TBX4	98,1	95,4	100	99,9	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX5	100	100	100	100	Holt-Oram syndrome, 142900
TBX6	99,2	94,8	100	100	Spondylocostal dysostosis 5, 122600
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TCF12	99,9	99,7	100	100	Craniosynostosis 3, 615314
TCIRG1	98,5	93,4	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCOF1	99,7	98,7	100	100	Treacher Collins syndrome 1, 154500
TCTN2	99,9	99,1	100	100	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100	100	100	100	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TENT5A	100	99,5	100	100	Osteogenesis imperfecta, type XVIII, 617952
TGDS	99,4	95,9	100	99,9	Catel-Manzke syndrome, 616145
TGFB1	100	98,8	100	100	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
TGFB2	100	100	100	100	Loeys-Dietz syndrome 4, 614816
TGFB3	100	100	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	93,6	93,6	98,8	97,6	Loeys-Dietz syndrome 1, 609192
TGFBR2	100	99,9	100	100	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
THPO	81,4	78,7	100	100	Thrombocythemia 1, 187950
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	98,5	92,8	100	100	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	100	99,3	100	100	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM251	100	98,8	100	100	Dysostosis multiplex, Ain-Naz type, 619345
TMEM38B	99,8	99,8	100	100	Osteogenesis imperfecta, type XIV, 615066
TMEM67	98,6	93,5	100	99,6	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688

					Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TNFRSF11A	94,9	93,8	99,1	97,7	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF11B	100	100	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	100	100	100	100	Osteopetrosis, autosomal recessive 2, 259710
TONSL	99,9	98,4	100	100	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TP63	100	100	100	100	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	98,7	95,4	100	100	Senior-Loken syndrome 9, 616629
TRAIP	100	100	100	99,9	Seckel syndrome 9, 616777
TRAPPC2	91,6	73,2	100	99,9	Spondyloepiphyseal dysplasia tarda, 313400
TREM2	100	99,3	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TRIP11	97,2	92,6	100	99,9	Odontochondrodysplasia 1, 184260 Achondrogenesis, type IA, 200600
TRPS1	100	99,9	100	100	Trichorhinophalangeal syndrome, type III, 190351 Trichorhinophalangeal syndrome, type I, 190350
TRPV4	100	99,9	100	100	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Digital arthropathy-brachydactyly, familial, 606835 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 Neuronopathy, distal hereditary motor, type VIII, 600175 Parastremmatic dwarfism, 168400 Brachyolmia type 3, 113500
TRPV6	100	99,9	99,9	99,2	Hyperparathyroidism, transient neonatal, 618188

TTC21B	99,7	99,1	100	99,9	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTI2	100	99,9	100	100	Mental retardation, autosomal recessive 39, 615541
TWIST1	100	99,4	96,7	90,6	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 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	99,7	98,9	100	99,9	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
VAC14	99,8	98,5	100	100	Striatonigral degeneration, childhood-onset, 617054
VDR	96,7	94,4	99,5	97,7	Rickets, vitamin D-resistant, type IIA, 277440
VPS33A	91,9	89,9	89,9	89,9	Mucopolysaccharidosis-plus syndrome, 617303
VPS35L	100	99,8	100	100	Ritscher-Schinzel syndrome 3, 619135
WDR19	99,8	98,6	100	99,9	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR35	99,6	98,4	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WNT1	99,9	96,6	100	100	Osteogenesis imperfecta, type XV, 615220
WNT10B	100	99,6	100	100	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300
WNT3	100	99,9	100	100	?Tetra-amelia syndrome 1, 273395
WNT5A	100	100	100	100	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100	99,3	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	99,7	98,4	100	100	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,8	91,1	97,7	94,1	Desbuquois dysplasia 2, 615777
XYLT2	99,9	97,1	96,7	96,7	Spondyloocular syndrome, 605822
ZBTB16	100	100	100	100	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type,

ZC4H2	100	98,1	100	100	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	99,6	99,4	100	99,9	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZSWIM6	95,1	91,6	94,3	91	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.
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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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors