SHORT STATURE AND SKELETAL DYSPLASIA GENE PANEL
DG 2.18 (521 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ABCC9	100%	99,90%	100%	100%	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ACAN	96,50%	92,70%	98,90%	98,70%	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	99,80%	98,30%	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVR1	100%	100%	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	99,90%	98,50%	100%	100%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	92,80%	89,00%	97,60%	95,80%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	97,10%	93,30%	99,80%	99,40%	Geleophysic dysplasia 1, 231050
AGA	100%	100%	100%	100%	Aspartylglucosaminuria, 208400
AGPS	99,30%	95,40%	100%	99,90%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AIFM1	99,90%	98,80%	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%	99,70%	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG9	100%	99,70%	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
ALPL	100%	100%	100%	100%	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300

					Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALX1	99,70%	97,10%	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	77,90%	73,30%	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	99,30%	100%	100%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMER1	99,90%	98,50%	100%	100%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100%	99,10%	100%	100%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKH	100%	100%	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	97,50%	94,80%	100%	100%	KBG syndrome, 148050
ANO5	99,50%	97,30%	100%	100%	Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307
ANTXR2	100%	98,20%	100%	100%	Hyaline fibromatosis syndrome, 228600
APC2	97,60%	92,70%	99,90%	99,10%	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
ARHGAP31	99,90%	98,80%	100%	100%	Adams-Oliver syndrome 1, 100300
ARID1B	99,50%	98,60%	99,90%	99,20%	Coffin-Siris syndrome 1, 135900
ARSB	96,90%	88,30%	100%	100%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSE	99,00%	93,00%	100%	99,90%	Chondrodysplasia punctata, X-linked recessive, 302950
ATP6V0A2	100%	99,50%	100%	100%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATR	99,90%	99,40%	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
B3GALT6	75,70%	69,70%	89,80%	81,60%	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	99,90%	98,20%	94,80%	94,80%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	99,80%	97,40%	99,90%	98,60%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BGN	100%	100%	100%	100%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	70,90%	50,40%	99,80%	97,30%	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 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877

BMPER	100%	99,80%	100%	100%	Diaphanospondylodysostosis, 608022
BMPR1B	100%	99,90%	100%	100%	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
BRAF	95,60%	85,10%	100%	100%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRF1	99,90%	98,40%	100%	100%	Cerebellofaciodental syndrome, 616202
BTK	100%	99,90%	100%	99,90%	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 Agammaglobulinemia, X-linked 1, 300755
BTRC	97,60%	97,30%	100%	100%	No OMIM disease ID
C16orf62	100%	99,90%	100%	100%	No OMIM disease ID
C21orf2	100%	99,30%	100%	100%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
C5orf42	99,70%	98,40%	100%	100%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	100%	100%	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CANT1	100%	99,90%	100%	100%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CASR	100%	99,90%	100%	100%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CBL	97,30%	97,10%	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CC2D2A	99,70%	97,70%	98,20%	98,20%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCDC8	100%	100%	100%	100%	3-M syndrome 3, 614205
CDC42	97,80%	90,70%	100%	100%	Takenouchi-Kosaki syndrome, 616737
CDC45	99,80%	98,50%	100%	100%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	100%	100%	?Meier-Gorlin syndrome 5, 613805
CDC73	100%	99,40%	100%	100%	Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001

					Parathyroid carcinoma, 608266 Hyperparathyroidism, familial primary, 145000
CDKN1C	86,30%	74,80%	99,20%	96,90%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDT1	99,70%	97,50%	100%	99,10%	Meier-Gorlin syndrome 4, 613804
CENPE	98,20%	92,20%	100%	100%	?Microcephaly 13, primary, autosomal recessive, 616051
CEP120	100%	99,50%	100%	100%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP152	99,70%	98,20%	100%	100%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	96,10%	90,00%	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
CHST14	99,90%	98,90%	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	99,40%	100%	100%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHSY1	97,20%	95,70%	99,70%	98,00%	Temtamy preaxial brachydactyly syndrome, 605282
CKAP2L	99,70%	98,60%	100%	100%	Filippi syndrome, 272440
CLCN5	99,90%	98,30%	100%	100%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
CLCN7	99,70%	98,40%	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%	99,90%	100%	100%	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COL10A1	100%	98,40%	100%	100%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	99,20%	95,70%	100%	100%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
COL11A2	100%	99,50%	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	99,90%	98,60%	100%	100%	Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL1A2	99,40%	97,00%	100%	100%	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	99,90%	99,70%	100%	100%	Steel syndrome, 615155
COL2A1	100%	99,70%	100%	100%	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 SED congenita, 183900 Legg-Calve-Perthes disease, 150600 SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL9A1	100%	99,20%	100%	100%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,90%	99,00%	100%	100%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	98,70%	95,50%	99,70%	98,60%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
COLEC11	100%	100%	100%	100%	3MC syndrome 2, 265050
COMP	93,40%	92,30%	100%	100%	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
CREB3L1	100%	99,90%	100%	100%	Osteogenesis imperfecta, type XVI, 616229

CREBBP	99,70%	98,50%	100%	100%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CRIPT	98,10%	93,20%	100%	100%	Short stature with microcephaly and distinctive facies, 615789
CRTAP	99,80%	98,80%	100%	100%	Osteogenesis imperfecta, type VII, 610682
CSF1R	99,90%	99,30%	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSGALNACT1	100%	99,80%	100%	100%	No OMIM disease ID
CTSA	100%	100%	100%	100%	Galactosialidosis, 256540
CTSK	100%	99,90%	100%	100%	Pycnodysostosis, 265800
CUL7	100%	99,30%	100%	100%	3-M syndrome 1, 273750
CYP26B1	100%	99,90%	100%	100%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	99,90%	99,30%	100%	100%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	99,40%	95,60%	100%	100%	Rickets due to defect in vitamin D 25-hydroxylation, 600081
DDR2	100%	99,90%	100%	100%	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100%	99,90%	100%	100%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX58	99,90%	99,00%	100%	100%	Singleton-Merten syndrome 2, 616298
DHCR24	100%	100%	100%	100%	Desmosterolosis, 602398
DHODH	100%	100%	100%	100%	Miller syndrome, 263750
DLL3	92,10%	87,00%	100%	99,10%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100%	99,20%	100%	100%	Adams-Oliver syndrome 6, 616589
DLX3	99,90%	98,40%	100%	100%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX5	100%	99,90%	100%	100%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	100%	100%	100%	100%	No OMIM disease ID
DMP1	100%	99,90%	100%	100%	Hypophosphatemic rickets, AR, 241520
DNA2	99,80%	98,30%	100%	100%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJC21	99,90%	99,00%	100%	100%	Bone marrow failure syndrome 3, 617052
DNMT3A	99,80%	98,60%	100%	100%	Heyn-Sproul-Jackson syndrome, 618724 Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DOCK6	99,30%	98,90%	100%	100%	Adams-Oliver syndrome 2, 614219
DONSON	91,70%	85,30%	100%	100%	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPCD	100%	100%	100%	100%	No OMIM disease ID

DPM1	98,20%	91,30%	99,70%	97,10%	Congenital disorder of glycosylation, type Ie, 608799
DSE	99,00%	96,10%	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DVL1	97,20%	95,00%	100%	100%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100%	100%	100%	100%	Robinow syndrome, autosomal dominant 3, 616894
DYM	97,40%	96,50%	100%	100%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC2H1	98,80%	95,50%	100%	100%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	99,70%	97,60%	100%	100%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EBP	99,70%	95,80%	100%	100%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EDN1	100%	100%	100%	100%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798
EDNRA	100%	100%	100%	100%	Mandibulofacial dysostosis with alopecia, 616367
EFL1	99,60%	98,50%	100%	100%	Shwachman-Diamond syndrome 2, 617941
EFNB1	100%	100%	100%	100%	Craniofrontonasal dysplasia, 304110
EFTUD2	100%	99,80%	100%	100%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2AK3	97,20%	94,50%	100%	100%	Wolcott-Rallison syndrome, 226980
EIF4A3	100%	99,50%	100%	100%	Robin sequence with cleft mandible and limb anomalies, 268305
ENPP1	96,40%	91,20%	98,70%	97,80%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
EOGT	79,40%	78,40%	91,90%	89,00%	Adams-Oliver syndrome 4, 615297
EP300	99,80%	99,00%	100%	100%	Rubinstein-Taybi syndrome 2, 613684 Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500
ERF	99,90%	98,50%	100%	100%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ESCO2	98,70%	95,20%	100%	100%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
EVC	93,90%	88,60%	96,90%	94,80%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	97,70%	96,10%	100%	100%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EXOC6B	99,10%	97,60%	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXT1	99,90%	98,40%	100%	100%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100%	99,30%	100%	100%	Exostoses, multiple, type 2, 133701 Seizures, scoliosis, and macrocephaly syndrome, 616682

EXTL3	100%	100%	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	100%	99,50%	100%	100%	Weaver syndrome, 277590
FAM111A	99,90%	99,30%	100%	100%	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM20B	100%	99,90%	100%	100%	No OMIM disease ID
FAM20C	100%	100%	100%	99,80%	Raine syndrome, 259775
FAM46A	100%	99,70%	100%	100%	Osteogenesis imperfecta, type XVIII, 617952
FAM58A	83,10%	78,50%	98,90%	94,70%	STAR syndrome, 300707
FAR1	97,60%	92,80%	100%	100%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBLN1	99,70%	97,60%	100%	99,70%	No OMIM disease ID
FBN1	100%	99,90%	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%	99,90%	100%	100%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBXW4	100%	99,40%	100%	100%	No OMIM disease ID
FERMT3	100%	100%	100%	100%	Leukocyte adhesion deficiency, type III, 612840
FGD1	97,30%	92,80%	100%	100%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	100%	99,80%	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	99,60%	97,50%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF8	98,20%	88,90%	100%	99,60%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	100%	100%	Multiple synostoses syndrome 3, 612961
FGFR1	100%	99,90%	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 HEARTsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001

FGFR2	97,70%	97,10%	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	99,80%	97,70%	100%	99,80%	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%	99,80%	100%	100%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FKBP10	98,80%	97,20%	100%	100%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	100%	99,90%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLNA	100%	99,90%	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	99,50%	98,80%	100%	100%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FMN1	99,60%	98,50%	100%	100%	No OMIM disease ID
FN1	100%	99,30%	100%	100%	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FUCA1	100%	99,90%	100%	100%	Fucosidosis, 230000
FUZ	100%	100%	100%	100%	No OMIM disease ID
FZD2	99,90%	98,20%	100%	100%	Omodysplasia 2, 164745
GALNS	100%	99,80%	100%	100%	Mucopolysaccharidosis IVA, 253000
GALNT3	99,80%	99,00%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GCM2	100%	100%	100%	100%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
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 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017
GDF6	100%	99,90%	100%	99,40%	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	99,60%	99,50%	99,80%	99,80%	Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 Growth hormone insensitivity, partial, 604271
GHRHR	96,40%	96,10%	100%	100%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	98,50%	95,80%	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	99,90%	97,40%	100%	100%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLI2	99,10%	97,40%	100%	99,80%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	100%	99,50%	100%	100%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GMNN	99,80%	97,40%	100%	100%	Meier-Gorlin syndrome 6, 616835
GNAI3	99,30%	95,20%	100%	100%	Auriculocondylar syndrome 1, 602483
GNAS	100%	99,90%	100%	99,90%	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	99,70%	97,30%	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765

GNPTAB	100%	99,90%	100%	100%	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	99,10%	94,30%	100%	99,90%	Mucolipidosis III gamma, 252605
GNS	98,40%	94,80%	100%	100%	Mucopolysaccharidosis type IIID, 252940
GORAB	100%	99,10%	100%	100%	Geroderma osteodysplasticum, 231070
GPC3	99,10%	94,70%	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	90,50%	85,80%	98,20%	94,90%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GSC	99,20%	92,40%	100%	100%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GUSB	92,90%	91,70%	100%	100%	Mucopolysaccharidosis VII, 253220
GZF1	100%	99,60%	100%	100%	Joint laxity, short stature, and myopia, 617662
HAAO	100%	99,80%	100%	100%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HDAC4	100%	99,80%	100%	100%	No OMIM disease ID
HDAC8	100%	99,80%	100%	100%	Cornelia de Lange syndrome 5, 300882
HES7	84,40%	53,90%	100%	100%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	99,70%	97,30%	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	86,40%	86,30%	91,20%	89,30%	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HMGA2	81,30%	76,70%	75,10%	73,80%	No OMIM disease ID
HOXA11	97,10%	87,50%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	77,70%	69,00%	89,70%	79,70%	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXD13	99,90%	98,60%	100%	100%	Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 ?Brachydactyly-syndactyly syndrome, 610713 Syndactyly, type V, 186300 Synpolydactyly 1, 186000
HPGD	100%	98,90%	100%	100%	Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HRAS	100%	100%	100%	100%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470

					Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HSPA9	88,50%	84,50%	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPG2	99,20%	97,70%	100%	99,90%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	100%	100%	100%	100%	Hydrolethalus syndrome, 236680
IARS2	100%	99,90%	100%	100%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
ICK	99,90%	98,70%	100%	100%	Endocrine-cerebroosteodysplasia, 612651
ID4	87,60%	82,50%	98,90%	93,10%	No OMIM disease ID
IDH1	93,30%	80,10%	100%	100%	No OMIM disease ID
IDH2	99,70%	97,40%	100%	99,80%	D-2-hydroxyglutaric aciduria 2, 613657
IDS	99,90%	98,00%	100%	100%	Mucopolysaccharidosis II, 309900
IDUA	93,70%	86,80%	100%	100%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IFIH1	99,70%	98,40%	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	99,30%	95,60%	100%	100%	Osteogenesis imperfecta, type V, 610967
IFT122	100%	99,60%	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	99,80%	98,80%	100%	100%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,90%	99,10%	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%	99,90%	100%	100%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	97,60%	88,20%	100%	100%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	93,50%	90,10%	95,00%	94,90%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100%	99,90%	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100%	99,90%	100%	100%	Insulin-like growth factor I, resistance to, 270450
IGF2	100%	100%	100%	100%	?Growth restriction, severe, with distinctive facies, 616489
IGFALS	99,90%	99,60%	100%	100%	Acid-labile subunit, deficiency of, 615961
IGSF1	99,50%	96,30%	100%	100%	Hypothyroidism, central, and testicular enlargement, 300888

IHH	100%	100%	100%	100%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	99,70%	96,50%	97,20%	97,20%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	84,10%	77,20%	100%	100%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IL1RN	100%	100%	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL2RG	99,80%	97,10%	100%	100%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL6ST	96,40%	90,30%	100%	100%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IMPAD1	100%	100%	100%	100%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INPPL1	98,40%	94,50%	99,90%	99,70%	Opsismodysplasia, 258480
INTU	99,70%	98,10%	100%	100%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
KAT6B	99,90%	99,00%	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
KIAA0586	97,30%	93,10%	95,80%	95,80%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	100%	99,30%	100%	100%	?Orofaciodigital syndrome XV, 617127
KIF22	100%	100%	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF7	93,60%	90,60%	99,10%	97,80%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KL	98,20%	97,20%	98,50%	97,50%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KMT2A	100%	99,90%	99,90%	99,40%	Wiedemann-Steiner syndrome, 605130
KRAS	99,50%	96,90%	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	99,40%	94,50%	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	99,90%	98,70%	100%	100%	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
LFNG	87,90%	86,40%	92,20%	87,70%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	96,60%	96,50%	100%	100%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100%	100%	100%	100%	Pituitary hormone deficiency, combined, 4, 262700
LIFR	99,70%	98,00%	100%	100%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMNA	97,40%	91,90%	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	99,60%	96,30%	100%	100%	Nail-patella syndrome, 161200
LONP1	100%	99,80%	100%	100%	CODAS syndrome, 600373
LPIN2	100%	100%	100%	100%	Majeed syndrome, 609628
LRP4	99,10%	98,80%	100%	100%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	98,50%	98,10%	100%	99,70%	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750

					Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634
LRRK1	98,60%	97,50%	100%	100%	No OMIM disease ID
LTBP2	99,90%	99,00%	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	99,60%	98,10%	100%	100%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LZTR1	100%	99,90%	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAFB	100%	99,40%	100%	100%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAN2B1	99,80%	97,90%	100%	100%	Mannosidosis, alpha-, types I and II, 248500
MANBA	99,80%	98,40%	100%	100%	Mannosidosis, beta, 248510
MAP2K1	99,80%	97,10%	100%	100%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	98,50%	95,10%	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K20	100%	99,50%	100%	100%	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAP3K7	100%	99,60%	100%	100%	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MATN3	84,70%	84,60%	100%	100%	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBTPS2	100%	99,00%	100%	100%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MECOM	100%	99,90%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MEGF8	99,90%	99,00%	100%	100%	Carpenter syndrome 2, 614976
MEOX1	100%	98,90%	100%	100%	Klippel-Feil syndrome 2, 214300
MESD	100%	99,90%	100%	100%	Osteogenesis imperfecta, type XX, 618644
MESP2	93,90%	86,90%	97,50%	97,50%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100%	99,50%	100%	100%	Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MGP	98,70%	95,10%	100%	100%	Keutel syndrome, 245150

MIR140	NC	NC	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MKS1	99,80%	97,90%	100%	100%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MMP13	95,20%	92,20%	92,40%	92,40%	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	100%	98,90%	100%	100%	?Winchester syndrome, 277950
MMP2	100%	100%	100%	100%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	99,10%	96,10%	100%	100%	Metaphyseal anadysplasia 2, 613073
MNX1	68,20%	58,30%	87,40%	79,20%	Currarino syndrome, 176450
MSX2	100%	99,40%	100%	100%	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550
MTAP	99,10%	93,50%	100%	100%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYCN	100%	99,90%	99,30%	96,70%	Feingold syndrome 1, 164280
MYH3	99,90%	99,00%	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
MYO18B	100%	99,10%	100%	100%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
NAGLU	92,90%	89,90%	99,90%	99,20%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NANS	100%	99,90%	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	100%	99,60%	100%	100%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NEK1	99,80%	98,00%	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	100%	99,60%	100%	100%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEU1	99,70%	97,70%	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NF1	92,60%	90,20%	100%	100%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210

NFIX	100%	99,50%	99,60%	98,70%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NIN	100%	99,50%	100%	100%	?Seckel syndrome 7, 614851
NIPBL	98,90%	97,00%	100%	100%	Cornelia de Lange syndrome 1, 122470
NKX3-2	99,80%	97,00%	100%	100%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP3	100%	99,90%	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	99,20%	97,20%	100%	100%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	100%	99,50%	100%	100%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPPC	100%	99,00%	100%	100%	No OMIM disease ID
NPR2	100%	99,60%	100%	100%	Short stature with nonspecific skeletal abnormalities, 616255 Epiphyseal chondrodysplasia, Miura type, 615923 Acromesomelic dysplasia, Maroteaux type, 602875
NPR3	100%	100%	100%	100%	No OMIM disease ID
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%	99,90%	100%	100%	Sotos syndrome 1, 117550
NSDHL	100%	98,70%	100%	100%	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	99,70%	98,20%	100%	100%	Seckel syndrome 10, 617253
NXN	100%	100%	99,90%	99,50%	Robinow syndrome, autosomal recessive 2, 618529
OBSL1	100%	99,30%	100%	100%	3-M syndrome 2, 612921

OFD1	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
ORC1	100%	99,40%	100%	100%	Meier-Gorlin syndrome 1, 224690
ORC4	98,70%	93,60%	100%	100%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	99,90%	100%	100%	Meier-Gorlin syndrome 3, 613803
OSTM1	98,60%	94,00%	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTX2	100%	99,70%	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	94,60%	94,00%	100%	100%	Cole-Carpenter syndrome 1, 112240
PAM16	65,30%	65,20%	82,90%	82,90%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	100%	99,90%	100%	100%	No OMIM disease ID
PAPSS2	100%	99,50%	100%	100%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PAX3	100%	99,90%	100%	100%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PCNT	99,60%	97,10%	100%	100%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	98,90%	95,50%	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE3A	99,90%	99,40%	100%	100%	Hypertension and brachydactyly syndrome, 112410
PDE4D	95,70%	93,50%	100%	99,80%	Acrodysostosis 2, with or without hormone resistance, 614613
PEX5	99,90%	99,00%	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,50%	86,70%	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	100%	99,60%	99,90%	99,20%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	99,90%	98,80%	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
TAB2	100%	99,70%	100%	100%	Congenital heart defects, nonsyndromic, 2, 614980
TAPT1	91,70%	86,90%	98,50%	94,80%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
PIGV	100%	100%	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300

PIK3R1	99,80%	99,00%	100%	100%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PISD	100%	100%	100%	100%	No OMIM disease ID
PITX1	96,70%	92,00%	100%	100%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	99,90%	97,70%	100%	100%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PKDCC	90,60%	81,50%	97,80%	94,70%	Rhizomelic limb shortening with dysmorphic features, 618821
PLCB3	100%	99,00%	100%	100%	No OMIM disease ID
PLCB4	99,90%	98,80%	100%	100%	Auriculocondylar syndrome 2, 614669
PLEKHM1	100%	99,80%	100%	100%	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLK4	99,90%	98,20%	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100%	98,40%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,30%	97,30%	100%	100%	Bruck syndrome 2, 609220
PLS3	97,70%	96,10%	97,20%	97,20%	Bone mineral density QTL18, osteoporosis, 300910
POC1A	100%	100%	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POLE	100%	99,80%	100%	100%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLL	100%	99,20%	100%	100%	No OMIM disease ID
POLR1A	100%	99,40%	100%	100%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	99,30%	95,50%	90,70%	90,70%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,60%	91,60%	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	100%	99,70%	100%	100%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,90%	98,60%	100%	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POP1	100%	99,70%	100%	100%	Anauxetic dysplasia 2, 617396
POR	99,80%	98,60%	100%	100%	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
POU1F1	100%	99,20%	100%	100%	Pituitary hormone deficiency, combined, 1, 613038
PPIB	100%	100%	100%	100%	Osteogenesis imperfecta, type IX, 259440
PPP1CB	99,90%	99,30%	100%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PRKAR1A	99,30%	93,50%	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
PROKR2	100%	100%	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	92,60%	82,60%	100%	100%	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	95,30%	81,60%	100%	100%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PTDSS1	100%	100%	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	100%	98,70%	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTHLH	99,70%	98,40%	100%	100%	Brachydactyly, type E2, 613382
PTPN11	99,10%	93,70%	100%	100%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PYCR1	99,90%	97,70%	100%	100%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	100%	99,50%	100%	100%	Carpenter syndrome, 201000
RAB33B	100%	100%	100%	100%	Smith-McCort dysplasia 2, 615222
RAC3	97,30%	94,40%	99,70%	98,20%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	99,20%	96,60%	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	99,70%	97,30%	100%	100%	?Bleeding disorder, platelet-type, 18, 615888
RBBP8	100%	99,70%	100%	100%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBM8A	99,80%	97,90%	100%	100%	Thrombocytopenia-absent radius syndrome, 274000
RBPJ	98,40%	92,80%	100%	100%	Adams-Oliver syndrome 3, 614814
RECQL4	99,80%	98,10%	100%	99,90%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400

RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RIPPLY2	100%	97,90%	100%	100%	?Spondylocostal dysostosis 6, 616566
RIT1	100%	100%	100%	100%	Noonan syndrome 8, 615355
RNPC3	91,50%	70,70%	100%	100%	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	NC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROR2	100%	99,90%	97,00%	97,00%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPL10	97,40%	89,10%	100%	100%	Mental retardation, X-linked, syndromic, 35, 300998
RPL13	96,30%	85,50%	100%	100%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RRAS	99,80%	95,70%	100%	100%	No OMIM disease ID
RSPO2	97,10%	90,70%	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	72,20%	72,20%	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	99,90%	99,00%	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	98,60%	96,70%	100%	100%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SBDS	100%	100%	100%	100%	Shwachman-Diamond syndrome, 260400
SCARF2	95,40%	86,30%	99,80%	99,20%	Van den Ende-Gupta syndrome, 600920
SEC24D	100%	99,70%	100%	100%	Cole-Carpenter syndrome 2, 616294
SERPINF1	100%	100%	100%	100%	Osteogenesis imperfecta, type VI, 613982
SERPINH1	100%	98,30%	100%	100%	Osteogenesis imperfecta, type X, 613848
SETD2	100%	99,90%	100%	100%	Luscan-Lumish syndrome, 616831
SF3B4	99,90%	97,30%	100%	100%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100%	99,80%	100%	100%	Pyle disease, 265900
SGMS2	100%	100%	100%	100%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGSH	94,40%	94,10%	100%	100%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3BP2	91,40%	91,20%	97,00%	95,30%	Cherubism, 118400

SH3PXD2B	100%	100%	100%	100%	Frank-ter Haar syndrome, 249420
SHH	100%	99,50%	100%	100%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	99,90%	99,40%	100%	100%	Noonan syndrome-like with loose anagen hair, 607721
SHOX	70,00%	59,70%	95,10%	95,10%	Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300
SKI	99,30%	94,90%	100%	99,40%	Shprintzen-Goldberg syndrome, 182212
SLC10A7	99,70%	98,00%	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC17A5	99,60%	97,00%	100%	100%	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC25A24	99,40%	99,30%	99,80%	99,80%	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%	99,60%	100%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC34A3	100%	99,40%	100%	100%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35D1	100%	97,70%	100%	100%	Schneckenbecken dysplasia, 269250
SLC39A13	99,80%	98,20%	100%	100%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLCO2A1	100%	99,40%	100%	100%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	99,70%	98,80%	100%	100%	No OMIM disease ID
SMAD2	100%	99,60%	100%	100%	No OMIM disease ID
SMAD3	99,90%	99,00%	100%	100%	Loeys-Dietz syndrome 3, 613795
SMAD4	100%	99,90%	100%	100%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA4	99,90%	99,00%	100%	100%	Coffin-Siris syndrome 4, 614609
SMARCAL1	100%	99,90%	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCB1	100%	100%	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCE1	96,10%	88,10%	100%	100%	Coffin-Siris syndrome 5, 616938

SMC1A	100%	98,70%	100%	99,80%	Cornelia de Lange syndrome 2, 300590 Epileptic encephalopathy, early infantile, 85, with or without midline brain defects, 301044
SMC3	95,20%	91,00%	100%	100%	Cornelia de Lange syndrome 3, 610759
SNRPB	100%	99,30%	100%	100%	Cerebrocostomandibular syndrome, 117650
SNX10	96,20%	95,70%	100%	99,60%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	99,80%	98,40%	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100%	99,20%	100%	100%	Noonan syndrome 9, 616559
SOST	100%	99,50%	100%	100%	Sclerosteosis 1, 269500 Van Buchem disease, 239100 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	91,40%	75,20%	100%	99,50%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX9	100%	98,60%	100%	100%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP7	100%	99,80%	100%	100%	Osteogenesis imperfecta, type XII, 613849
SPARC	100%	100%	100%	100%	Osteogenesis imperfecta, type XVII, 616507
SPECC1L	100%	99,60%	100%	100%	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SPINK5	99,90%	99,50%	100%	100%	Netherton syndrome, 256500
SPR	99,80%	96,30%	100%	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100%	98,90%	100%	100%	Legius syndrome, 611431
SRCAP	99,40%	98,90%	100%	100%	Floating-Harbor syndrome, 136140
SRP54	99,50%	96,50%	100%	100%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
STAT3	100%	99,80%	100%	100%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	100%	98,50%	100%	100%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
SULF1	99,90%	99,30%	100%	100%	No OMIM disease ID
SUMF1	97,50%	90,80%	100%	100%	Multiple sulfatase deficiency, 272200
TBCE	99,80%	97,30%	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBX15	100%	99,90%	100%	100%	Cousin syndrome, 260660

TBX3	99,20%	96,80%	100%	100%	Ulnar-mammary syndrome, 181450
TBX4	97,60%	95,10%	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	99,50%	95,40%	100%	100%	Spondylocostal dysostosis 5, 122600
TBXAS1	100%	100%	100%	100%	Ghosal hematodiaphyseal syndrome, 231095
TCF12	100%	99,90%	100%	100%	Craniosynostosis 3, 615314
TCIRG1	97,60%	90,10%	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TCOF1	99,70%	98,60%	100%	100%	Treacher Collins syndrome 1, 154500
TCTEX1D2	100%	100%	100%	100%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN2	100%	99,50%	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TGDS	99,40%	96,80%	100%	100%	Catel-Manzke syndrome, 616145
TGFB1	100%	99,90%	100%	100%	Camurati-Engelmann disease, 131300 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	93,70%	93,60%	99,00%	96,30%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	100%	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
THPO	100%	99,50%	100%	100%	Thrombocythemia 1, 187950
TMEM165	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	99,90%	98,10%	100%	100%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100%	99,60%	100%	100%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM38B	100%	99,90%	100%	100%	Osteogenesis imperfecta, type XIV, 615066
TMEM67	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TNFRSF11A	94,60%	93,30%	99,20%	98,00%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301

TNFRSF11B	100%	100%	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	100%	99,90%	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TONSL	99,80%	97,80%	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	99,60%	97,60%	100%	100%	Senior-Loken syndrome 9, 616629
TRAIP	100%	100%	100%	100%	Seckel syndrome 9, 616777
TRAPPC2	89,70%	69,60%	100%	100%	Spondyloepiphyseal dysplasia tarda, 313400
TREM2	100%	99,80%	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TRIP11	98,40%	94,00%	100%	100%	Osteochondrodysplasia, 184260 Achondrogenesis, type IA, 200600
TRPS1	100%	99,90%	100%	100%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV4	100%	99,90%	100%	100%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 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%	99,50%	99,90%	98,90%	Hyperparathyroidism, transient neonatal, 618188
TTC21B	99,90%	99,30%	100%	100%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TWIST1	100%	98,90%	97,20%	92,30%	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%	99,60%	100%	100%	?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 ?Hip dysplasia, Beukes type, 142669

VAC14	99,90%	98,50%	100%	100%	Striatonigral degeneration, childhood-onset, 617054
VDR	99,90%	99,00%	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440
VPS33A	97,30%	95,70%	95,80%	95,80%	Mucopolysaccharidosis-plus syndrome, 617303
WDR19	100%	99,40%	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%	99,60%	100%	100%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	99,80%	98,90%	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	99,50%	97,00%	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WISP3	100%	100%	100%	100%	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 Arthropathy, progressive pseudorheumatoid, of childhood, 208230
WNT1	99,30%	95,30%	100%	100%	Osteogenesis imperfecta, type XV, 615220
WNT10B	100%	99,40%	100%	100%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT3	100%	99,60%	100%	100%	?Tetra-amelia syndrome 1, 273395
WNT5A	100%	100%	100%	100%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100%	98,70%	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	99,90%	99,30%	100%	100%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,40%	89,60%	98,10%	94,80%	Desbuquois dysplasia 2, 615777
XYLT2	100%	98,30%	96,70%	96,70%	Spondyloocular syndrome, 605822
ZBTB16	100%	99,90%	100%	100%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZMPSTE24	100%	99,90%	100%	100%	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZSWIM6	95,50%	91,90%	94,90%	92,10%	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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors