SHORT STATURE AND SKELETAL DYSPLASIA GENE PANEL
DG 3.5.0 (579 genes)

Releasedate: 05-12-2022

Gene	TWIST X2 covered >10x	TWIST X2 covered >20x	Associated Phenotype description and OMIM disease ID
ABCC9	100%	100%	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 ?Atrial fibrillation, familial, 12, 614050 Intellectual disability and myopathy syndrome, 619719
ACAN	99%	99%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACP5	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	100%	100%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACVR1	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ADAMTS10	100%	100%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	100%	100%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTSL2	100%	100%	Geleophysic dysplasia 1, 231050
AFF3	100%	100%	KINSSHIP syndrome, 619297
AGA	100%	100%	Aspartylglucosaminuria, 208400
AGPS	100%	100%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AIFM1	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%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG3	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG9	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	100%	100%	Alstrom syndrome, 203800

ALPL	100%	100%	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
ALX1	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMER1	100%	100%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100%	100%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKH	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKRD11	100%	100%	KBG syndrome, 148050
ANO5	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260
ANTXR2	100%	100%	Hyaline fibromatosis syndrome, 228600
APC2	100%	100%	Cortical dysplasia, complex, with other brain malformations 10, 618677 Intellectual developmental disorder, autosomal recessive 74, 617169
ARCN1	100%	100%	Short stature-micrognathia syndrome, 617164
ARHGAP31	100%	100%	Adams-Oliver syndrome 1, 100300
ARID1B	99%	98%	Coffin-Siris syndrome 1, 135900
ARSB	100%	100%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSL	100%	100%	Chondrodysplasia punctata, X-linked recessive, 302950
ATP6V0A2	100%	100%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATR	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
B3GALT6	100%	98%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GAT3	95%	94%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	100%	100%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BGN	100%	100%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	100%	100%	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432

BMP1	100%	100%	Osteogenesis imperfecta, type XIII, 614856
BMP2	100%	100%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BMPER	100%	100%	Diaphanospondylodysostosis, 608022
BMPR1B	100%	100%	Acromesomelic dysplasia 3, 609441 Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849
IMPAD1	100%	100%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BRAF	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	100%	100%	Cerebellofaciodental syndrome, 616202
BTK	100%	100%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	100%	100%	No OMIM disease ID
BUB1B	100%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
CA2	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CANT1	100%	100%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CASR	100%	100%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980
CBL	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CC2D2A	98%	98%	COACH syndrome 2, 619111 Retinitis pigmentosa 93, 619845 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCDC134	100%	100%	Osteogenesis imperfecta, type XXII, 619795
CCDC8	100%	100%	3-M syndrome 3, 614205
CCN6	100%	100%	Progressive pseudorheumatoid dysplasia, 208230
CCNQ	100%	100%	STAR syndrome, 300707

CDC42	100%	100%	Takenouchi-Kosaki syndrome, 616737
CDC45	100%	100%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	?Meier-Gorlin syndrome 5, 613805
CDC73	100%	100%	Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism-jaw tumor syndrome, 145001
CDH3	100%	100%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDKN1C	100%	100%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDT1	100%	100%	Meier-Gorlin syndrome 4, 613804
CENPE	100%	100%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPJ	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP120	100%	100%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP152	100%	100%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	100%	100%	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP57	100%	100%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	100%	100%	?Seckel syndrome 6, 614728
CFAP410	100%	100%	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
CHST11	100%	100%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	100%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHSY1	100%	100%	Temtamy preaxial brachydactyly syndrome, 605282
CILK1	100%	100%	Endocrine-cerebroosteodysplasia, 612651
CKAP2L	100%	100%	Filippi syndrome, 272440
CLCN5	100%	100%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Hypophosphatemic rickets, 300554

			Dent disease 1, 300009 Nephrolithiasis, type I, 310468
CLCN7	100%	100%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
COG1	100%	100%	Congenital disorder of glycosylation, type IIg, 611209
COG4	100%	100%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COL10A1	100%	100%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	100%	100%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	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	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	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	100%	100%	Steel syndrome, 615155
COL2A1	100%	100%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 Czech dysplasia, 609162 Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 SMED Strudwick type, 184250 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450

			SED congenita, 183900 Kniest dysplasia, 156550 Stickler syndrome, type I, nonsyndromic ocular, 609508 Osteoarthritis with mild chondrodysplasia, 604864 Stickler syndrome, type I, 108300 Platyspondylic skeletal dysplasia, Torrance type, 151210 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600
COL9A1	100%	100%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COL9A3	100%	100%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 Stickler syndrome, type VI, 620022
COLEC11	100%	100%	3MC syndrome 2, 265050
COMP	100%	100%	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400
CPLANE1	100%	100%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CREB3L1	100%	100%	Osteogenesis imperfecta, type XVI, 616229
CREBBP	100%	100%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRIPT	100%	100%	Short stature with microcephaly and distinctive facies, 615789
CRTAP	100%	100%	Osteogenesis imperfecta, type VII, 610682
CSF1R	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSGALNACT1	100%	100%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CTSA	100%	100%	Galactosialidosis, 256540
CTSK	100%	100%	Pycnodysostosis, 265800
CUL7	100%	100%	3-M syndrome 1, 273750
CYP26B1	100%	100%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP27B1	100%	100%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	100%	100%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
DDR2	100%	100%	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100%	100%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557

DHCR24	100%	100%	Desmosterolosis, 602398
DHODH	100%	100%	Miller syndrome, 263750
DLL3	100%	100%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100%	100%	Adams-Oliver syndrome 6, 616589
DLX3	100%	100%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX5	100%	100%	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	100%	100%	No OMIM disease ID
DMP1	100%	100%	Hypophosphatemic rickets, AR, 241520
DNA2	100%	100%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJC21	100%	100%	Bone marrow failure syndrome 3, 617052
DNMT3A	100%	100%	Tatton-Brown-Rahman syndrome, 615879 Acute myeloid leukemia, somatic, 601626 Heyn-Sproul-Jackson syndrome, 618724
DOCK6	100%	100%	Adams-Oliver syndrome 2, 614219
DONSON	100%	100%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPCD	100%	100%	No OMIM disease ID
DPF2	100%	100%	Coffin-Siris syndrome 7, 618027
DPM1	99%	97%	Congenital disorder of glycosylation, type Ie, 608799
DSE	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DVL1	100%	100%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100%	100%	Robinow syndrome, autosomal dominant 3, 616894
DYM	100%	100%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC2H1	100%	99%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	100%	100%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	100%	100%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	100%	100%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EBP	100%	100%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECEL1	100%	100%	Arthrogryposis, distal, type 5D, 615065
EDN1	100%	100%	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706

EDNRA	100%	100%	Mandibulofacial dysostosis with alopecia, 616367
EFL1	100%	100%	Shwachman-Diamond syndrome 2, 617941
EFNB1	100%	100%	Craniofrontonasal dysplasia, 304110
EFTUD2	100%	100%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2AK3	100%	100%	Wolcott-Rallison syndrome, 226980
EIF4A3	100%	100%	Robin sequence with cleft mandible and limb anomalies, 268305
ENPP1	100%	100%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
EOGT	98%	94%	Adams-Oliver syndrome 4, 615297
EP300	100%	100%	Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
ERF	100%	100%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ESCO2	100%	100%	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
EVC	100%	100%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	100%	100%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC6B	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC5	100%	100%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXT1	100%	100%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100%	100%	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	100%	100%	Weaver syndrome, 277590
FAM111A	100%	100%	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361
FAM20B	100%	100%	No OMIM disease ID
FAM20C	100%	100%	Raine syndrome, 259775
FAR1	100%	100%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FBLN1	100%	100%	No OMIM disease ID

FBN1	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%	100%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FBXW4	100%	100%	No OMIM disease ID
FERMT3	100%	100%	Leukocyte adhesion deficiency, type III, 612840
FGD1	100%	100%	Intellectual developmental disorder, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	100%	99%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF8	100%	100%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	Multiple synostoses syndrome 3, 612961
FGFR1	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	100%	100%	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 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 ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500

			Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,
FGFR3	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	100%	100%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FKBP10	100%	100%	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450
FKBP14	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLNA	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	100%	100%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721

			Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FMN1	100%	100%	No OMIM disease ID
FN1	100%	100%	Spondylometaphyseal dysplasia, corner fracture type, 184255 Glomerulopathy with fibronectin deposits 2, 601894
FUCA1	100%	100%	Fucosidosis, 230000
FUZ	100%	100%	No OMIM disease ID
FZD2	100%	100%	Omodysplasia 2, 164745
GALNS	100%	100%	Mucopolysaccharidosis IVA, 253000
GALNT2	100%	100%	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GCM2	100%	100%	Hypoparathyroidism, familial isolated 2, 618883 Hyperparathyroidism 4, 617343
GDF3	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia, isolated, with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	100%	100%	Acromesomelic dysplasia 2A, 200700 Acromesomelic dysplasia 2B, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A2, 112600 ?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250 Brachydactyly, type C, 113100 Brachydactyly, type A1, C, 615072
GDF6	100%	100%	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%	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	100%	100%	Laron dwarfism, 262500 Increased responsiveness to growth hormone, 604271 Growth hormone insensitivity, partial, 604271
GHRHR	100%	100%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	100%	100%	Growth hormone deficiency, isolated partial, 615925

GINS2	100%	100%	No OMIM disease ID
GJA1	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	100%	100%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLI2	100%	100%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	100%	100%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GMNN	100%	100%	Meier-Gorlin syndrome 6, 616835
GNAI3	100%	100%	Auriculocondylar syndrome 1, 602483
GNAS	100%	100%	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	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	100%	100%	?Rhizomelic dysplasia, Ain-Naz type, 616510
GNPTAB	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	100%	100%	Mucolipidosis III gamma, 252605
GNS	100%	100%	Mucopolysaccharidosis type IIID, 252940
GORAB	100%	100%	Geroderma osteodysplasticum, 231070
GPC3	100%	99%	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870

GPC6	100%	100%	Omodysplasia 1, 258315
GPR161	100%	100%	No OMIM disease ID
GPX4	100%	100%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GSC	100%	100%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GUSB	100%	100%	Mucopolysaccharidosis VII, 253220
GZF1	100%	100%	Joint laxity, short stature, and myopia, 617662
H19	NC	NC	No OMIM disease ID
HAAO	100%	100%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HDAC4	100%	100%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC8	98%	97%	Cornelia de Lange syndrome 5, 300882
HES7	100%	100%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HGSNAT	92%	92%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HMGA2	90%	81%	Silver-Russell syndrome 5, 618908
HOXA11	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	100%	99%	Hand-foot-uterus syndrome, 140000 ?Guttmacher syndrome, 176305
HOXD13	100%	100%	Syndactyly, type V, 186300 Synpolydactyly 1, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 ?Brachydactyly-syndactyly syndrome, 610713
HPGD	100%	100%	?Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HRAS	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
HS2ST1	100%	100%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HSPA9	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170

HSPG2	100%	100%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HYLS1	100%	100%	Hydrolethalus syndrome, 236680
IARS2	100%	100%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
ID4	100%	100%	No OMIM disease ID
IDH1	100%	100%	No OMIM disease ID
IDH2	100%	100%	D-2-hydroxyglutaric aciduria 2, 613657
IDS	100%	100%	Mucopolysaccharidosis II, 309900
IDUA	100%	100%	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IFIH1	100%	100%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	100%	100%	Osteogenesis imperfecta, type V, 610967
IFT122	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	100%	100%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	100%	100%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100%	100%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100%	100%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	100%	100%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	95%	95%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100%	100%	Insulin-like growth factor I, resistance to, 270450
IGF2	100%	100%	Silver-Russell syndrome 3, 616489
IGFALS	100%	100%	Acid-labile subunit, deficiency of, 615961
IGSF1	100%	100%	Hypothyroidism, central, and testicular enlargement, 300888
IHH	100%	100%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	100%	100%	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204

IKBKG	100%	98%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636 Autoinflammatory disease, systemic, X-linked, 301081
IL1RN	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL2RG	100%	100%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL6ST	100%	100%	Stuve-Wiedemann syndrome 2, 619751 Hyper-IgE recurrent infection syndrome 4A, autosomal dominant, 619752 ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 Hyper-IgE recurrent infection syndrome 4B, autosomal recessive, 618523
INPPL1	100%	100%	Opsismodysplasia, 258480
INTU	100%	100%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
KAT6B	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNJ2	100%	100%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KDELR2	100%	100%	Osteogenesis imperfecta, type XXI, 619131
KIAA0586	96%	96%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA0753	100%	100%	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIF22	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF24	100%	100%	No OMIM disease ID
KIF7	100%	100%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KL	100%	99%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KMT2A	100%	100%	Wiedemann-Steiner syndrome, 605130
KRAS	100%	100%	Gastric cancer, somatic, 613659 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470

			Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
KYNU	100%	100%	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
LAMA5	100%	100%	Nephrotic syndrome, type 26, 620049 ?Bent bone dysplasia syndrome 2, 620076
LBR	100%	100%	Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471 Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 Greenberg skeletal dysplasia, 215140
LBX1	100%	100%	?Central hypoventilation syndrome, congenital, 3, 619483
LEMD3	100%	100%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700
LFNG	99%	97%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LHX3	100%	100%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100%	100%	Pituitary hormone deficiency, combined, 4, 262700
LIFR	100%	100%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LMBR1	100%	99%	Triphalangeal thumb, type I, 174500 Syndactyly, type IV, 186200 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500 Acheiropody, 200500 Triphalangeal thumb-polysyndactyly syndrome, 190605
LMNA	100%	100%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Restrictive dermopathy 2, 619793 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660

			Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LMX1B	100%	100%	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LONP1	100%	100%	CODAS syndrome, 600373
LPIN2	100%	100%	Majeed syndrome, 609628
LRP4	100%	100%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	100%	100%	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	100%	100%	Osteosclerotic metaphyseal dysplasia, 615198
LTBP1	100%	100%	Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	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	100%	100%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
TMEM251	100%	100%	Dysostosis multiplex, Ain-Naz type, 619345
LZTR1	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAFB	100%	100%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAN2B1	100%	100%	Mannosidosis, alpha-, types I and II, 248500
MANBA	100%	100%	Mannosidosis, beta, 248510
MAP2K1	100%	100%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K20	100%	100%	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	100%	100%	Frontometaphyseal dysplasia 2, 617137 Cardiospondylocarpofacial syndrome, 157800
MAPK1	100%	100%	Noonan syndrome 13, 619087

MATN3	100%	100%	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBTPS2	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%	100%	?Meier-Gorlin syndrome 8, 617564
MECOM	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MEGF8	100%	100%	Carpenter syndrome 2, 614976
MEOX1	100%	100%	Klippel-Feil syndrome 2, 214300
MESD	100%	100%	Osteogenesis imperfecta, type XX, 618644
MESP2	100%	100%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100%	100%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 ?Arthrogryposis, distal, type 11, 620019 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705
MGP	100%	100%	Keutel syndrome, 245150
MIR140	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MKS1	100%	100%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MMP13	92%	92%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400
MMP14	100%	100%	?Winchester syndrome, 277950
MMP2	100%	100%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP9	100%	100%	Metaphyseal anadysplasia 2, 613073
MNX1	98%	93%	Currarino syndrome, 176450
MRAS	100%	100%	Noonan syndrome 11, 618499
MSX2	100%	100%	Parietal foramina with cleidocranial dysplasia, 168550 Craniosynostosis 2, 604757 Parietal foramina 1, 168500
MTAP	100%	100%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MYCN	100%	100%	Feingold syndrome 1, 164280
MYH3	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%	Arthrogryposis, distal, type 1C, 619110
MYO18B	100%	100%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
NADSYN1	100%	100%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGLU	100%	100%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NANS	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBAS	100%	100%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NEK1	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK9	100%	100%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022
NEU1	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NF1	100%	100%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFIX	100%	100%	Marshall-Smith syndrome, 602535 Malan syndrome, 614753
NIN	100%	100%	?Seckel syndrome 7, 614851
NIPBL	100%	100%	Cornelia de Lange syndrome 1, 122470
NKX3-2	100%	100%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NLRP3	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
NMNAT1	100%	98%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553
NOG	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	100%	100%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	100%	100%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPPC	100%	100%	No OMIM disease ID
NPR2	100%	100%	Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255 Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100%	100%	Boudin-Mortier syndrome, 619543
NRAS	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%	100%	Sotos syndrome, 117550
NSD2	100%	100%	Rauch-Steindl syndrome, 619695
NSDHL	100%	100%	CK syndrome, 300831 CHILD syndrome, 308050
NSMCE2	100%	100%	Seckel syndrome 10, 617253
NXN	100%	100%	Robinow syndrome, autosomal recessive 2, 618529
OBSL1	100%	100%	3-M syndrome 2, 612921
OFD1	100%	100%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
ORC1	100%	100%	Meier-Gorlin syndrome 1, 224690
ORC4	99%	98%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	100%	Meier-Gorlin syndrome 3, 613803
OSTM1	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTX2	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%	Osteogenesis imperfecta, type VIII, 610915

P4HB	100%	100%	Cole-Carpenter syndrome 1, 112240
PAM16	85%	85%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAPPA2	100%	100%	Short stature, Dauber-Argente type, 619489
PAPSS2	100%	100%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PAX3	100%	100%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220
PCNT	100%	100%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT1A	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE3A	100%	100%	Hypertension and brachydactyly syndrome, 112410
PDE4D	100%	100%	Acrodysostosis 2, with or without hormone resistance, 614613
PEX5	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	91%	91%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHEX	100%	99%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGV	100%	100%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PIK3R1	100%	100%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PISD	100%	100%	Liberfarb syndrome, 618889
PITX1	100%	100%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	100%	100%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PKDCC	100%	100%	Rhizomelic limb shortening with dysmorphic features, 618821
PLAG1	100%	100%	Adenomas, salivary gland pleomorphic, somatic, 181030 Silver-Russell syndrome 4, 618907
PLCB3	100%	100%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	100%	100%	Auriculocondylar syndrome 2, 614669

PLEKHM1	100%	100%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLK4	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100%	100%	Bruck syndrome 2, 609220
PLOD3	100%	100%	Lysyl hydroxylase 3 deficiency, 612394
PLS3	97%	97%	Bone mineral density QTL18, osteoporosis, 300910
PNPLA6	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
POC1A	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POLE	100%	100%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLL	100%	100%	No OMIM disease ID
POLR1A	100%	100%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	83%	83%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	100%	100%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100%	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POP1	100%	100%	Anauxetic dysplasia 2, 617396
POR	100%	100%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
POU1F1	100%	100%	Pituitary hormone deficiency, combined or isolated, 1, 613038
PPIB	100%	100%	Osteogenesis imperfecta, type IX, 259440
PPM1D	100%	100%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPP1CB	100%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R21	100%	100%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PRKACA	100%	100%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 Cardioacrofacial dysplasia 1, 619142
PRKACB	100%	99%	Cardioacrofacial dysplasia 2, 619143
PRKAR1A	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	100%	100%	Spondylometaphyseal dysplasia, Pagnamenta type, 619638 Acromesomelic dysplasia 4, 619636
PROKR2	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100%	100%	Pituitary hormone deficiency, combined, 2, 262600
PSAT1	100%	100%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMB1	100%	100%	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038
PTDSS1	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTH1R	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
PTHLH	100%	100%	Brachydactyly, type E2, 613382
PTPN11	100%	100%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PUF60	100%	100%	Verheij syndrome, 615583
PYCR1	100%	100%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	100%	100%	Carpenter syndrome, 201000
RAB33B	100%	100%	Smith-McCort dysplasia 2, 615222
RAC3	100%	100%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	100%	100%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RAF1	100%	100%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RALA	100%	100%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RASGRP2	100%	100%	?Bleeding disorder, platelet-type, 18, 615888
RBBP8	100%	100%	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,
RBM8A	100%	100%	Thrombocytopenia-absent radius syndrome, 274000

RBPJ	100%	100%	Adams-Oliver syndrome 3, 614814
RECQL4	100%	100%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
DDX58	100%	100%	Singleton-Merten syndrome 2, 616298
RIPPLY2	100%	100%	?Spondylocostal dysostosis 6, 616566
RIT1	100%	100%	Noonan syndrome 8, 615355
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNPC3	100%	100%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU4ATAC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROR2	100%	100%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	100%	100%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPL10	100%	100%	Intellectual developmental disorder, X-linked syndromic 35, 300998
RPL13	100%	100%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RRAS	100%	100%	No OMIM disease ID
RRAS2	100%	100%	Noonan syndrome 12, 618624 Ovarian carcinoma,
RREB1	100%	100%	No OMIM disease ID
RSPO2	100%	100%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPRY1	100%	100%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RUNX2	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	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	100%	100%	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SATB2	100%	100%	Glass syndrome, 612313

SBDS	100%	100%	Shwachman-Diamond syndrome 1, 260400
SCARF2	100%	100%	Van den Ende-Gupta syndrome, 600920
SCUBE3	100%	100%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184
SEC24D	100%	100%	Cole-Carpenter syndrome 2, 616294
SEMA3A	100%	100%	No OMIM disease ID
SERPINF1	100%	100%	Osteogenesis imperfecta, type VI, 613982
SERPINH1	100%	100%	Osteogenesis imperfecta, type X, 613848
SETD2	100%	100%	Luscan-Lumish syndrome, 616831
SF3B4	100%	100%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100%	100%	Pyle disease, 265900
SGMS2	100%	100%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGSH	100%	100%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3BP2	100%	99%	Cherubism, 118400
SH3PXD2B	100%	100%	Frank-ter Haar syndrome, 249420
SHH	100%	100%	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHOC2	100%	100%	Noonan syndrome-like with loose anagen hair 1, 607721
SHOX	95%	95%	Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300 Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300
SKI	100%	100%	Shprintzen-Goldberg syndrome, 182212
SLC10A7	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC17A5	100%	100%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC25A24	100%	100%	Fontaine progeroid syndrome, 612289
SLC26A2	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%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782

SLC34A3	100%	100%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35C1	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	100%	Schneckenbecken dysplasia, 269250
SLC39A13	100%	100%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC4A2	100%	100%	No OMIM disease ID
SLCO2A1	100%	100%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	100%	100%	No OMIM disease ID
SMAD2	100%	100%	Loeys-Dietz syndrome 6, 619656 Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	100%	100%	Loeys-Dietz syndrome 3, 613795
SMAD4	100%	100%	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA4	100%	100%	Coffin-Siris syndrome 4, 614609
SMARCA5	100%	100%	No OMIM disease ID
SMARCAL1	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCB1	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCE1	100%	100%	Coffin-Siris syndrome 5, 616938
SMC1A	100%	100%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	Cornelia de Lange syndrome 3, 610759
SMOC2	100%	100%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNRPB	100%	100%	Cerebrocostomandibular syndrome, 117650
SNX10	100%	100%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100%	100%	Noonan syndrome 9, 616559
SOST	100%	100%	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX2	100%	100%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SOX3	100%	100%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000

SOX9	100%	100%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SP7	100%	100%	Osteogenesis imperfecta, type XII, 613849
SPARC	100%	100%	Osteogenesis imperfecta, type XVII, 616507
SPECC1L	100%	100%	Teebi hypertelorism syndrome 1, 145420 ?Facial clefting, oblique, 1, 600251
SPINK5	100%	100%	Netherton syndrome, 256500
SPR	100%	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100%	100%	Legius syndrome, 611431
SPRED2	100%	100%	Noonan syndrome 14, 619745
SRCAP	100%	100%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 Floating-Harbor syndrome, 136140
SRP54	100%	100%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
STAT3	100%	100%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	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	100%	100%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
SULF1	100%	100%	No OMIM disease ID
SUMF1	100%	100%	Multiple sulfatase deficiency, 272200
TAB2	100%	100%	Congenital heart defects, nonsyndromic, 2, 614980
TAPT1	100%	100%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBCE	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBX15	100%	99%	Cousin syndrome, 260660
TBX2	100%	100%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX3	100%	100%	Ulnar-mammary syndrome, 181450
TBX4	100%	100%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX5	100%	100%	Holt-Oram syndrome, 142900
TBX6	100%	100%	Spondylocostal dysostosis 5, 122600
TBXAS1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095

TCF12	100%	100%	Craniosynostosis 3, 615314 Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCIRG1	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TCOF1	100%	100%	Treacher Collins syndrome 1, 154500
TCTN2	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TENT5A	100%	100%	Osteogenesis imperfecta, type XVIII, 617952
TGDS	100%	100%	Catel-Manzke syndrome, 616145
TGFB1	100%	100%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
TGFB2	100%	100%	Loeys-Dietz syndrome 4, 614816
TGFB3	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	100%	100%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
THPO	100%	100%	Thrombocythemia 1, 187950
TMEM165	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	100%	100%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	100%	100%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM38B	100%	100%	Osteogenesis imperfecta, type XIV, 615066
TMEM67	100%	98%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TNFRSF11A	100%	100%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF11B	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFSF11	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TONSL	100%	100%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510

TP63	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	100%	100%	Senior-Loken syndrome 9, 616629
TRAIP	100%	100%	Seckel syndrome 9, 616777
TRAPPC2	100%	100%	Spondyloepiphyseal dysplasia tarda, 313400
TREM2	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TRIM37	98%	98%	Mulibrey nanism, 253250
TRIP11	100%	100%	Odontochondrodysplasia 1, 184260 Achondrogenesis, type IA, 200600
TRIP13	100%	100%	Oocyte maturation defect 9, 619011 Mosaic variegated aneuploidy syndrome 3, 617598
TRPS1	100%	100%	Trichorhinophalangeal syndrome, type III, 190351 Trichorhinophalangeal syndrome, type I, 190350
TRPV4	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%	100%	Hyperparathyroidism, transient neonatal, 618188
TTC21B	100%	100%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTI2	100%	100%	Intellectual developmental disorder, autosomal recessive 39, 615541
TWIST1	100%	100%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TYROBP	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770

UFSP2	100%	100%	?Hip dysplasia, Beukes type, 142669 Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 Developmental and epileptic encephalopathy 106, 620028
VAC14	100%	100%	Striatonigral degeneration, childhood-onset, 617054
VDR	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440
VPS33A	90%	90%	Mucopolysaccharidosis-plus syndrome, 617303
VPS35L	100%	100%	Ritscher-Schinzel syndrome 3, 619135
WDR19	100%	100%	Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378 Senior-Loken syndrome 8, 616307 Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Spermatogenic failure 72, 619867
WDR35	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WNT1	100%	100%	Osteogenesis imperfecta, type XV, 615220
WNT10B	100%	100%	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300
WNT3	100%	100%	?Tetra-amelia syndrome 1, 273395
WNT5A	100%	100%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100%	100%	No OMIM disease ID
WNT7A	100%	100%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
XRCC4	100%	100%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	100%	100%	Desbuquois dysplasia 2, 615777
XYLT2	100%	99%	Spondyloocular syndrome, 605822
ZBTB16	100%	100%	Leukemia, acute promyelocytic, PL2F/RARA type,
ZC4H2	100%	100%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy 1, 275210
ZSWIM6	98%	96%	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.
Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 is the chemistry used for 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 : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors