Whole Exome Sequencing
Gene package Skeletal Dysplasia, Version 1, 8-3-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ABCC9	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850	601439	55	100	98	90
ACAN	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	155760	221	100	100	98
ACP5	Spondyloenchondrodysplasia with immune dysregulation, 607944	171640	133	100	100	100
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	208	100	100	100
ACVR1	Fibrodysplasia ossificans progressiva, 135100	102576	65	100	100	96
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600	608990	103	100	100	99
ADAMTS17	Weill-Marchesani 4 syndrome, recessive, 613195	607511	96	100	96	93
ADAMTSL2	Geleophysic dysplasia 1, 231050	612277	89	100	100	97
AGA	Aspartylglucosaminuria, 208400	613228	62	100	99	92
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121	603051	56	100	97	82
ALG12	Congenital disorder of glycosylation, type Ig, 607143	607144	152	100	100	100
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	78	100	100	100

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ALG9	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210	606941	54	100	99	89
ALMS1	Alstrom syndrome, 203800	606844	84	100	99	97
ALPL	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300	171760	113	100	100	99
AMER1	Osteopathia striata with cranial sclerosis, 300373	300647	62	100	99	95
ANKH	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000	605145	79	100	100	99
ANKRD11	KBG syndrome, 148050	611192	111	100	100	98
ANO5	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, type 2L, 611307	608662	65	100	99	92
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	611542	68	100	100	95
ARSE	Chondrodysplasia punctata, X-linked recessive, 302950	300180	69	100	97	83
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640	615291	66	100	75	72
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600	606374	125	100	100	100
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070	604327	129	100	100	98
BMP1	Osteogenesis imperfecta, type XIII, 614856	112264	107	100	100	100
BMPER	Diaphanospondylodysostosis, 608022	608699	83	100	100	94
BMPR1B	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849 Brachydactyly, type A2, 112600	603248	61	100	100	98
BRAF	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706	164757	60	100	99	90
BTK	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755	300300	41	100	96	68
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730	611492	80	100	100	99
CANT1	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719	613165	124	100	100	100

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CBL	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	69	100	100	99
CCDC8	3-M syndrome 3, 614205	614145	194	100	100	100
CCN6	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230	603400	83	86	77	69
CDC45	Meier-Gorlin syndrome 7, 617063	603465	88	100	99	96
CDC6	?Meier-Gorlin syndrome 5, 613805	602627	54	100	100	95
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	80	100	85	78
CDT1	Meier-Gorlin syndrome 4, 613804	605525	114	100	99	95
CEP120	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300	613446	63	100	99	92
CFAP410	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271	603191	44	100	96	74
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095	603799	120	100	100	100
CLCN5	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990	300008	51	100	99	91
CLCN7	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490	602727	119	100	100	99
COG1	Congenital disorder of glycosylation, type IIg, 611209	606973	89	100	99	95
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500	120110	84	100	100	99
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	53	100	98	84
COL11A2	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150	120290	96	100	100	98

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COL1A1	{Bone mineral density variation QTL, osteoporosis}, 166710 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220	120150	120	100	100	99
COL1A2	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 imperfecta, type III, 259420 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710	120160	62	100	99	94
COL2A1	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia	120140	92	100	100	98
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	57	100	97	87
COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	120260	99	100	100	96
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932	120270	89	100	97	92
COLEC11	3MC syndrome 2, 265050	612502	158	100	100	100
COMP	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170	600310	107	100	98	95
CREB3L1	No OMIM phenotype	616215	97	100	100	97

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CREBBP	Rubinstein-Taybi syndrome 1, 180849	600140	82	100	98	93
CRTAP	Osteogenesis imperfecta, type VII, 610682	605497	79	100	100	97
CSGALNACT1	No OMIM phenotype	616615	106	100	100	100
CTSA	Galactosialidosis, 256540	613111	118	100	100	100
CTSK	Pycnodysostosis, 265800	601105	51	100	100	95
CUL7	3-M syndrome 1, 273750	609577	100	100	100	99
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416	605207	158	100	100	100
CYP27B1	Vitamin D-dependent rickets, type I, 264700	609506	109	100	100	100
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665	191311	78	100	100	96
DHCR24	Desmosterolosis, 602398	606418	113	100	100	100
DLL3	Spondylocostal dysostosis 1, autosomal recessive, 277300	602768	95	100	97	91
DLX3	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320	600525	139	100	99	77
DMP1	Hypophosphatemic rickets, AR, 241520	600980	59	100	99	96
DONSON	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230	611428	39	100	91	71
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	70	100	89	83
DVL1	Robinow syndrome, autosomal dominant 2, 616331	601365	119	100	100	100
DYM	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326	607461	56	100	96	81
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	603297	61	100	97	84
EBP	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960	300205	76	100	100	100
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	62	100	99	91
ENPP1	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Obesity, susceptibility to}, 601665	173335	48	100	91	77
EVC	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530	604831	92	100	95	93
EVC2	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	607261	80	100	99	94
EXT1	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700	608177	63	100	98	92
EXT2	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682	608210	89	100	99	94
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425	605744	133	100	100	100

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FAM111A	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000	615292	62	100	100	99
FAM20C	Raine syndrome, 259775	611061	119	100	100	100
FBN1	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328	134797	175	100	100	100
FERMT3	Leukocyte adhesion deficiency, type III, 612840	607901	109	100	100	95
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400	300546	65	100	99	95
FGF23	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993	605380	86	100	100	96
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702	600483	133	100	95	94
FGF9	Multiple synostoses syndrome 3, 612961	600921	70	100	100	100
FGFR1	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440	136350	94	100	100	98

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FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly, maxillary retrusion, and mental retardation, 609579	176943	63	100	99	90
FGFR3	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601	134934	112	100	100	99
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	53	100	99	88
FKBP10	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968	607063	123	100	100	100

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FLNA	Cardiac valvular dysplasia, 314400 Congenital short bowel syndrome, 300048 ?FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	300017	98	100	100	99
FLNB	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460	603381	100	100	100	97
FN1	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255	135600	69	100	99	93
FUCA1	Fucosidosis, 230000	612280	79	100	100	94
FZD2	No OMIM phenotype	600667	131	100	98	97
GALNS	Mucopolysaccharidosis IVA, 253000	612222	76	100	98	93
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900	601756	60	100	100	95
GDF3	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704	606522	105	100	100	100
GDF5	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 {Osteoarthritis-5}, 612400 Symphalangism, proximal, 1B, 615298	601146	139	100	100	100

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GDF6	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898	601147	127	100	100	100
GH1	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650	139250	167	100	100	100
GHR	Growth hormone insensitivity, partial, 604271 {Hypercholesterolemia, familial, modifier of}, 143890 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500	600946	63	100	100	96
GHRHR	Growth hormone deficiency, isolated, type IB, 612781	139191	77	100	100	97
GHSR	Growth hormone deficiency, isolated partial, 615925	601898	118	100	100	100
GJA1	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100	121014	82	100	100	97
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	96	100	100	96
GLI2	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829	165230	144	100	100	98
GLI3	Greig cephalopolysyndactyly syndrome, 175700 {Hypothalamic hamartomas, somatic}, 241800 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700	165240	102	100	100	98
GMNN	Meier-Gorlin syndrome 6, 616835	602842	53	100	99	88

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GNAS	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463	139320	164	100	99	97
GNPAT	Rhizomelic chondrodysplasia punctata, type 2, 222765	602744	66	100	99	91
GNPTAB	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600	607840	54	100	98	89
GNPTG	Mucolipidosis III gamma, 252605	607838	149	100	100	95
GNS	Mucopolysaccharidosis type IIID, 252940	607664	60	100	100	96
GORAB	Geroderma osteodysplasticum, 231070	607983	55	100	100	95
GPC6	Omodysplasia 1, 258315	604404	69	100	100	96
GPX4	Spondylometaphyseal dysplasia, Sedaghatian type, 250220	138322	178	100	100	97
GUSB	Mucopolysaccharidosis VII, 253220	611499	100	100	100	97
HDAC4	No OMIM phenotype	605314	104	100	100	99
HES7	Spondylocostal dysostosis 4, autosomal recessive, 613686	608059	65	100	86	77
HESX1	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230	601802	60	100	100	88
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	60	100	94	88
HOXA13	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000	142959	79	100	79	75
HPGD	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100	601688	65	100	100	93
HRAS	{Bladder cancer, somatic}, 109800 Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470	190020	178	100	100	100
HSPA9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854	600548	62	100	98	90

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HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800	142461	102	100	99	98
HYLS1	Hydrolethalus syndrome, 236680	610693	61	100	100	100
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	99	100	100	99
IDS	Mucopolysaccharidosis II, 309900	300823	68	100	98	84
IDUA	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016	252800	127	100	99	94
IFITM5	Osteogenesis imperfecta, type V, 610967	614757	132	100	100	100
IFT122	Cranioectodermal dysplasia 1, 218330	606045	108	100	100	98
IFT140	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	110	100	99	97
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	65	100	99	92
IFT43	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866	614068	70	100	100	98
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	611177	55	100	96	80
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747	147440	63	100	100	99
IGF1R	Insulin-like growth factor I, resistance to, 270450	147370	108	100	100	99
IGFALS	Acid-labile subunit, deficiency of, 615961	601489	115	100	100	100
IGSF1	Hypothyroidism, central, and testicular enlargement, 300888	300137	51	100	97	87
IHH	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500	600726	129	100	100	100
IKBKB	Immunodeficiency 15, 615592	603258	79	100	99	95
IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640	300248	77	100	100	96
IL2RG	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400	308380	49	100	99	81
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type, 614078	614010	125	100	99	94
INPPL1	Opsismodysplasia, 258480	600829	101	100	100	99
KIAA0753	?Orofaciodigital syndrome XV, 617127	617112	45	100	96	83
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546	603213	144	100	100	100

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KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	104	100	96	93
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	60	100	100	95
KRAS	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	190070	73	100	97	72
LBR	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019 ?Reynolds syndrome, 613471	600024	57	100	99	92
LEMD3	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700	607844	84	100	99	92
LFNG	?Spondylocostal dysostosis 3, autosomal recessive, 609813	602576	112	100	83	82
LHX3	Pituitary hormone deficiency, combined, 3, 221750	600577	106	100	100	100
LHX4	Pituitary hormone deficiency, combined, 4, 262700	602146	87	100	100	100
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559	151443	52	100	96	81
LMX1B	Nail-patella syndrome, 161200	602575	112	100	100	100
LONP1	CODAS syndrome, 600373	605490	129	100	100	100
LRP4	Cenani-Lenz syndactyly syndrome, 212780 ?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305	604270	93	100	99	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
LRP5	[Bone mineral density variability 1], 601884 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 {Osteoporosis}, 166710 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636	603506	137	100	99	97
LRRK1	No OMIM phenotype	610986	101	99	95	90
LTBP2	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	602091	105	100	100	99
LTBP3	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809	602090	125	100	99	96
LZTR1	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670	600574	116	100	100	100
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	110	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	76	100	98	92
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	72	100	100	92
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	111	100	100	95
MAP3K7	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137	602614	50	100	99	88
MATN3	Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600 ?Spondyloepimetaphyseal dysplasia, 608728	602109	65	100	89	82
MBTPS2	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918	300294	39	100	96	79
MEOX1	Klippel-Feil syndrome 2, 214300	600147	59	100	100	86
MESP2	Spondylocostal dysostosis 2, autosomal recessive, 608681	605195	143	100	100	100
MGP	Keutel syndrome, 245150	154870	58	100	98	88
MMP13	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111	600108	69	100	99	91
MMP14	?Winchester syndrome, 277950	600754	132	100	100	100
MMP2	Multicentric osteolysis, nodulosis, and arthropathy, 259600	120360	106	100	100	99

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MMP9	Metaphyseal anadysplasia 2, 613073	120361	110	100	100	99
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250	156540	65	100	99	85
MYH3	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110	160720	86	100	100	96
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	114	100	97	93
NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442	605202	57	100	99	93
NBAS	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800	608025	57	100	98	89
NEK1	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520	604588	50	100	97	81
NEK9	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025	609798	55	100	97	84
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	143	100	100	100
NIN	?Seckel syndrome 7, 614851	608684	72	100	98	90
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330	602183	105	100	100	100
NOTCH2	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500	600275	92	100	100	98
NPPC	No OMIM phenotype	600296	104	100	100	100
NPR2	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255	108961	106	100	100	99
NRAS	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470	164790	53	100	100	95
OBSL1	3-M syndrome 2, 612921	610991	124	100	100	99
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	37	100	89	63

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ORC1	Meier-Gorlin syndrome 1, 224690	601902	76	100	99	93
ORC4	Meier-Gorlin syndrome 2, 613800	603056	57	100	98	79
ORC6	Meier-Gorlin syndrome 3, 613803	607213	65	100	100	92
OSTM1	Osteopetrosis, autosomal recessive 5, 259720	607649	69	100	99	92
OTX2	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125	600037	86	100	100	100
P3H1	Osteogenesis imperfecta, type VIII, 610915	610339	111	100	100	100
P4HB	Cole-Carpenter syndrome 1, 112240	176790	96	100	100	100
PAM16	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320	614336	88	100	100	100
PAPPA2	No OMIM phenotype	No ID	88	100	99	98
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847	603005	91	100	99	92
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	121	100	100	98
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940	123695	67	100	97	75
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799	600129	62	100	97	91
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	104	100	100	99
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	49	100	100	95
PHEX	Hypophosphatemic rickets, X-linked dominant, 307800	300550	39	100	96	78
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	135	100	100	99
PHYH	Refsum disease, 266500	602026	109	100	98	87
PIK3R1	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	72	100	100	94
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550	602149	99	100	100	96
PITX2	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550	601542	139	100	100	100
PLEKHM1	Osteopetrosis, autosomal recessive 6, 611497	611466	181	100	100	100
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171	605031	53	100	98	87
PLOD2	Bruck syndrome 2, 609220	601865	48	100	96	81
PLS3	Bone mineral density QTL18, osteoporosis, 300910	300131	41	100	92	74
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	78	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
POP1	Anauxetic dysplasia 2, 617396	602486	63	100	98	91
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	124015	152	100	100	100
POU1F1	Pituitary hormone deficiency, combined, 1, 613038	173110	68	100	100	99
PPIB	Osteogenesis imperfecta, type IX, 259440	123841	90	100	100	99
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489	188830	77	100	100	96
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200	607123	198	100	100	100
PROP1	Pituitary hormone deficiency, combined, 2, 262600	601538	104	100	100	93
PSAT1	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992	610936	57	100	100	91
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050	612792	55	100	100	93
PTH1R	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400	168468	120	100	100	98
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	66	100	98	84
RAB33B	Smith-McCort dysplasia 2, 615222	605950	69	100	100	100
RAF1	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553	164760	66	100	98	90
RASGRP2	?Bleeding disorder, platelet-type, 18, 615888	605577	90	100	100	100
RBM8A	Thrombocytopenia-absent radius syndrome, 274000	605313	81	100	100	100
RBPJ	Adams-Oliver syndrome 3, 614814	147183	64	100	96	83
RIPPLY2	?Spondylocostal dysostosis 6, 616566	609891	73	100	100	90
RIT1	Noonan syndrome 8, 615355	609591	62	100	100	98
RMRP	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460	157660	No coverage data			
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651	601428	59	95	81	65

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ROR2	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310	602337	122	100	100	99
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	52	100	94	81
RPL10	{Autism, susceptibility to, X-linked 5}, 300847 Mental retardation, X-linked, syndromic, 35, 300998	312173	89	100	100	100
RSPRY1	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723	616585	64	100	95	83
RUNX2	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510	600211	83	100	100	97
SBDS	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400	607444	72	100	100	98
SCARF2	Van den Ende-Gupta syndrome, 600920	613619	106	100	99	98
SEC24D	Cole-Carpenter syndrome 2, 616294	607186	65	100	99	91
SERPINF1	Osteogenesis imperfecta, type VI, 613982	172860	99	100	98	94
SERPINH1	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504	600943	142	100	100	100
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	102	100	95	89
SH3PXD2B	Frank-ter Haar syndrome, 249420	613293	116	100	100	98
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	51	100	98	89
SHOX	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582	312865	62	100	100	93
SLC10A7	No OMIM phenotype	611459	48	100	94	79
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	73	100	100	94
SLC25A24	Fontaine progeroid syndrome, 612289	608744	68	100	99	85
SLC26A2	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900	606718	59	100	100	99
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, 602782	612373	139	100	99	99
SLC34A3	Hypophosphatemic rickets with hypercalciuria, 241530	609826	120	100	95	91
SLC35D1	Schneckenbecken dysplasia, 269250	610804	51	100	97	77

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SLC39A13	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350	608735	124	100	100	100
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441	601460	94	100	100	98
SLCO5A1	No OMIM phenotype	613543	91	100	99	92
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	69	100	100	96
SMARCAL1	Schimke immunoosseous dysplasia, 242900	606622	77	100	100	96
SNRPB	Cerebrocostomandibular syndrome, 117650	182282	90	100	100	100
SNX10	Osteopetrosis, autosomal recessive 8, 615085	614780	65	100	100	94
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	62	100	99	89
SOS2	Noonan syndrome 9, 616559	601247	66	100	98	90
SOST	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100	605740	183	100	100	100
SOX2	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900	184429	161	100	100	100
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000	313430	66	100	96	92
SOX9	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290	608160	120	100	100	100
SP7	?Osteogenesis imperfecta, type XII, 613849	606633	127	100	100	100
SPARC	Osteogenesis imperfecta, type XVII, 616507	182120	84	100	98	96
SPINK5	Netherton syndrome, 256500	605010	55	100	98	89
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	100	100	100	100
SRCAP	Floating-Harbor syndrome, 136140	611421	116	100	100	99
STAT3	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060	102582	80	100	100	96
STAT5B	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578	604260	115	100	99	93
SULF1	No OMIM phenotype	610012	66	100	100	94
SUMF1	Multiple sulfatase deficiency, 272200	607939	83	100	100	96
TAPT1	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897	612758	36	98	87	63
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460	604934	53	100	95	82

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TBX15	Cousin syndrome, 260660	604127	68	100	100	96
TBX4	Ischiocoxopodopatellar syndrome, 147891	601719	128	100	100	100
TBX6	Spondylocostal dysostosis 5, 122600	602427	96	100	100	96
TBXAS1	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158	274180	68	100	98	90
TCIRG1	Osteopetrosis, autosomal recessive 1, 259700	604592	116	100	100	99
TCTEX1D2	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405	617353	81	100	100	86
TCTN2	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	613846	73	100	100	94
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	59	100	100	94
TGFB1	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700	190180	109	100	99	97
TMEM165	Congenital disorder of glycosylation, type IIk, 614727	614726	109	100	100	98
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613277	89	100	100	89
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	101	100	100	99
TMEM38B	Osteogenesis imperfecta, type XIV, 615066	611236	58	100	100	93
TNFRSF11A	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080	603499	90	100	95	95
TNFRSF11B	Paget disease of bone 5, juvenile-onset, 239000	602643	78	100	100	97
TNFSF11	Osteopetrosis, autosomal recessive 2, 259710	602642	47	100	99	84
TRAPPC2	Spondyloepiphyseal dysplasia tarda, 313400	300202	43	100	91	70
TRIP11	Achondrogenesis, type IA, 200600	604505	58	100	96	85
TRPS1	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351	604386	73	100	100	99

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TRPV4	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Scapuloperoneal spinal muscular atrophy, 181405 [Sodium serum level QTL 1], 613508 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252	605427	102	100	100	100
TTC21B	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819	612014	63	100	98	89
VDR	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440	601769	78	100	100	100
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	62	100	99	92
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	613363	109	100	100	100
WDR35	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	613602	52	100	98	86
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	615462	67	100	99	91
WNT1	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221	164820	195	100	100	100
WNT5A	Robinow syndrome, autosomal dominant 1, 180700	164975	113	100	100	97
XRCC4	Short stature, microcephaly, and endocrine dysfunction, 616541	194363	48	100	98	85
XYLT1	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822	608125	97	100	97	92
XYLT2	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800	608124	128	100	100	100
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447	176797	114	100	100	100
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210	606480	51	100	99	88

- Gene symbols according HGCN
- OMIM release used: 4-7-2018
- "No OMIM phenotypes" indicates a gene without a current OMIM association

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x

- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 95 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x