CHD panel

versie	V1 (471 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AATK	605276	No OMIM phenotype
ABL1	189980	Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant; Leukemia, Philadelphia chromosome-positive, resistant to imatinib (3)
ACAN	155760	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 (3), Autosomal recessive; Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3), Autosomal dominant
ACTA1	102610	Myopathy, actin, congenital, with cores, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal recessive, Autosomal dominant; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal recessive, Autosomal dominant
ACTB	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
ACTC1	102540	Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Left ventricular noncompaction 4, 613424 (3), Autosomal dominant
ACTG1	102560	Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant; Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant
ACTN2	102573	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant
ACVR1	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ACVR1B	601300	Pancreatic cancer, somatic (3)
ACVR2A	102581	No OMIM phenotype
ACVR2B	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
ACVRL1	601284	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant
ADAMTS10	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive

ADNP	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
AKAP12	No OMIM gene	No OMIM phenotype
AKT1	164730	Breast cancer, somatic, 114480 (3); Colorectal cancer, somatic, 114500 (3); Cowden syndrome 6, 615109 (3); Ovarian cancer, somatic, 167000 (3); Proteus syndrome, somatic, 176920 (3); {Schizophrenia, susceptibility to}, 181500 (2), Autosomal dominant
AKT2	164731	Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant
ALDH1A2	603687	No OMIM phenotype
ALX3	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
ANKRD1	609599	No OMIM phenotype
ANKRD11	611192	KBG syndrome, 148050 (3), Autosomal dominant
AP1B1	600157	No OMIM phenotype
APAF1	602233	No OMIM phenotype
AR	313700	Androgen insensitivity, 300068 (3), X-linked recessive; Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive
ARHGAP31	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARID4A	180201	No OMIM phenotype
ARMC4	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
ATP2A2	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATRX	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
AXIN1	603816	?Caudal duplication anomaly, 607864 (3); Hepatocellular carcinoma, somatic, 114550 (3)
BCOR	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BMP10	608748	No OMIM phenotype
BMP2	112261	Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive
BMP4	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
BMP6	112266	No OMIM phenotype
BMP7	112267	No OMIM phenotype
BMPR1A	601299	Juvenile polyposis syndrome, infantile form, 174900 (3), Autosomal dominant; Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant

BMPR2	600799	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 (3), Autosomal dominant; Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 (3), Autosomal dominant; Pulmonary venoocclusive disease 1, 265450 (3), Autosomal dominant
BRAF	164757	Adenocarcinoma of lung, somatic, 211980 (3); Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Nonsmall cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
BRIP1	605882	Breast cancer, early-onset, 114480 (3), Autosomal dominant; Fanconi anemia, complementation group J, 609054 (3)
BVES	604577	?Muscular dystrophy, limb-girdle, type 2X, 616812 (3), Autosomal recessive
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive
CAV1	601047	?Lipodystrophy, congenital generalized, type 3, 612526 (3); ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 (3), Autosomal dominant; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant
CAV3	601253	Cardiomyopathy, familial hypertrophic, 192600 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type IC, 607801 (3), Autosomal recessive, Autosomal dominant; Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Rippling muscle disease, 606072 (3), Autosomal dominant
CBL	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
CCDC103	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
CCDC114	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
CCDC151	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
CCDC39	613798	Ciliary dyskinesia, primary, 14, 613807 (3)
CCDC40	613799	Ciliary dyskinesia, primary, 15, 613808 (3)
CCDC65	611088	Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive
CCNO	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
CD96	606037	C syndrome, 211750 (3), Autosomal recessive
CDK13	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
CDK4	123829	{Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant
CDX2	600297	No OMIM phenotype
CENPF	600236	Stromme syndrome, 243605 (3), Autosomal recessive

CFAP53 (CCDC11)	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
CFC1	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
CHAC2	617446	No OMIM phenotype
CHD4	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD7	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
CHD8	610528	{Autism, susceptibility to, 18}, 615032 (3), Autosomal dominant
CITED2	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
COL1A1	120150	{Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, classic, 130000 (3), Autosomal dominant; Ehlers-Danlos syndrome, type VIIA, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant
COL1A2	120160	Ehlers-Danlos syndrome, cardiac valvular form, 225320 (3), Autosomal recessive; Ehlers-Danlos syndrome, type VIIB, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant
COL2A1	120140	Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Stickler sydrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)

COX7B	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
CREBBP	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRELD1	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
CRIP2	601183	No OMIM phenotype
CRYAB	123590	Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive
CSNK1D	600864	Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant
CSRP1	123876	No OMIM phenotype
CSRP3	600824	?Cardiomyopathy, dilated, 1M, 607482 (3); Cardiomyopathy, hypertrophic, 12, 612124 (3), Autosomal dominant
CTNNB1	116806	Colorectal cancer, somatic, 114500 (3); Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Medulloblastoma, somatic, 155255 (3); Mental retardation, autosomal dominant 19, 615075 (3), Autosomal dominant; Ovarian cancer, somatic, 167000 (3); Pilomatricoma, somatic, 132600 (3)
CYR61	602369	No OMIM phenotype
DDX3X	300160	Mental retardation, X-linked 102, 300958 (3), X-linked recessive, X-linked dominant
DES	125660	Cardiomyopathy, dilated, 1I, 604765 (3); ?Muscular dystrophy, limb-girdle, type 2R, 615325 (3), Autosomal recessive; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant; Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant
DIAPH3	614567	Auditory neuropathy, autosomal dominant, 1, 609129 (3), Autosomal dominant
DICER1	606241	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 (3), Autosomal dominant; Pleuropulmonary blastoma, 601200 (3), Autosomal dominant; Rhabdomyosarcoma, embryonal, 2, 180295 (3)
DLGAP5	No OMIM gene	No OMIM phenotype
DLL1	No OMIM gene	No OMIM phenotype
DLL4	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DNAAF1	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAAF2	612517	Ciliary dyskinesia, primary, 10, 612518 (3)
DNAAF3	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive

DNAAF4 (DYX1C1)	608706	Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive; {Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant
DNAAF5 (HEATR2)	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3)
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3)
DNAJB13	610263	Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive
DNAL1	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
DRC1	615288	Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
DSCAM	602523	No OMIM phenotype
DVL1	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
DYRK1A	600855	Mental retardation, autosomal dominant 7, 614104 (3), Autosomal dominant
E2F1	189971	No OMIM phenotype
ECE1	600423	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 (3), Autosomal dominant; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
EDNRA	131243	Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant; {Migraine, resistance to}, 157300 (3), Autosomal dominant
EFEMP2	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
EFNB1	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
EFTUD2	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EGFR	131550	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal recessive; ?Inflammatory skin and bowel disease, neonatal, 2, 616069 (3), Autosomal recessive; Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal recessive; {Nonsmall cell lung cancer, susceptibility to}, 211980 (3), Autosomal recessive
EGR1	128990	No OMIM phenotype
EHMT1	607001	Kleefstra syndrome, 610253 (3), Autosomal dominant
ELN	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
ENG	131195	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant
EOGT	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive

EP300	602700	Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EPHB2	600997	{Prostate cancer/brain cancer susceptibility, somatic}, 603688 (3)
EPHB4	600011	{Hydrops fetalis, nonimmune, and/or atrial septal defect}, 617300 (3), Autosomal dominant
ERBB2	164870	Adenocarcinoma of lung, somatic, 211980 (3); Gastric cancer, somatic, 613659 (3); Glioblastoma, somatic, 137800 (3); Ovarian cancer, somatic (3)
ERCC4	133520	Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; ?XFE progeroid syndrome, 610965 (3); Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive
ESR2	601663	No OMIM phenotype
ETS1	164720	No OMIM phenotype
EVC	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EWSR1	133450	Ewing sarcoma, 612219 (3); Neuroepithelioma, 612219 (3)
EZH2	601573	Weaver syndrome, 277590 (3), Autosomal dominant
F2R	187930	No OMIM phenotype
FABP3	134651	No OMIM phenotype
FADD	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759 (3), Autosomal recessive
FANCA	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
FANCB	300515	Fanconi anemia, complementation group B, 300514 (3)
FANCC	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
FANCD2	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FANCE	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
FANCF	613897	Fanconi anemia, complementation group F, 603467 (3)
FANCG	602956	Fanconi anemia, complementation group G, 614082 (3)
FANCI	611360	Fanconi anemia, complementation group I, 609053 (3)
FANCL	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
FBLN1	135820	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4), Autosomal dominant
FBLN7	611551	No OMIM phenotype

FBN1	134797	Acromicric dysplasia, 102370 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; MASS syndrome, 604308 (3); Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant
FBN2	612570	Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant; Macular degeneration, early-onset, 616118 (3), Autosomal dominant
FGF10	602115	Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant
FGF8	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGFR1	136350	Encephalocraniocutaneous lipomatosis, 613001 (3), Somatic mosaicism; Hartsfield syndrome, 615465 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant
FGFR2	176943	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal recessive; Apert syndrome, 101200 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Crouzon syndrome, 123500 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)

FGFR3	134934	Achondroplasia, 100800 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); SADDAN, 616482 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant
FHL2	602633	No OMIM phenotype
FIBP	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
FLI1	193067	Bleeding disorder, platelet-type, 21, 617443 (3)
FLNA	300017	Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked recessive; Congenital short bowel syndrome, 300048 (3), X-linked recessive; FG syndrome 2, 300321 (3); Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive; Heterotopia, periventricular, 300049 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3)
FLNC	102565	Cardiomyopathy, familial hypertrophic, 26 (3); Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant
FLT1	165070	No OMIM phenotype
FLT4	136352	Hemangioma, capillary infantile, somatic, 602089 (3); Lymphedema, hereditary, IA, 153100 (3), Autosomal dominant
FN1	135600	Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant; Plasma fibronectin deficiency, 614101 (1), Autosomal dominant
FOXC1	601090	Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant
FOXC2	602402	Lymphedema-distichiasis syndrome, 153400 (3), Autosomal dominant; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3), Autosomal dominant
FOXH1	603621	No OMIM phenotype
FOXK2	147685	No OMIM phenotype
FOXL1	603252	No OMIM phenotype

FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal dominant; Premature ovarian failure 3, 608996 (3), Autosomal dominant
FOXO1	136533	Rhabdomyosarcoma, alveolar, 268220 (3), Autosomal recessive
FOXP1	605515	Mental retardation with language impairment and with or without autistic features, 613670 (3), Autosomal dominant
FSCN2	607643	Retinitis pigmentosa 30, 607921 (3)
G6PC3	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
GAS8	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
GATA2	137295	Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
GATA4	600576	Atrial septal defect 2, 607941 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant
GATA5	611496	No OMIM phenotype
GATA6	601656	Atrial septal defect 9, 614475 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
GATAD2B	614998	Mental retardation, autosomal dominant 18, 615074 (3), Autosomal dominant
GDF1	602880	Double-outlet right ventricle, 217095 (3); Right atrial isomerism, 208530 (3), Autosomal recessive; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Transposition of great arteries, dextro-looped 3, 613854 (3), Autosomal dominant
GDF11	603936	No OMIM phenotype
GDF5	601146	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type C, 113100 (3), Autosomal dominant; Chondrodysplasia, Grebe type, 200700 (3), Autosomal recessive; Du Pan syndrome, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Symphalangism, proximal, 1B, 615298 (3)

GDNF	600837	Central hypoventilation syndrome, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant; {Pheochromocytoma, modifier of}, 171300 (3), Autosomal dominant
GJA1	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva 3, 617525 (3); Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
GJA5	121013	Atrial fibrillation, familial, 11, 614049 (3), Autosomal dominant; Atrial standstill, digenic (GJA5/SCN5A), 108770 (3), Autosomal dominant
GJA9	611923	No OMIM phenotype
GLB1	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive
GLI2	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
GLI3	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; {Hypothalamic hamartomas, somatic}, 241800 (3); Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
GPC5	602446	No OMIM phenotype
GPC6	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
GRK5	600870	No OMIM phenotype
GTF2I	601679	No OMIM phenotype
GTF2IRD1	604318	No OMIM phenotype
HAND1	No OMIM gene	No OMIM phenotype
HAND2	602407	No OMIM phenotype
HDAC8	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HES4	608060	No OMIM phenotype
HEY2	604674	No OMIM phenotype
HIF1A	603348	No OMIM phenotype
HMGB2	163906	No OMIM phenotype

HNF1A	142410	Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant; Hepatic adenoma, somatic, 142330 (3); MODY, type III, 600496 (3), Autosomal dominant; Renal cell carcinoma, 144700 (3)
HNF4A	600281	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant
HOXA1	142955	Athabaskan brainstem dysgenesis syndrome, 601536 (3); Bosley-Salih-Alorainy syndrome, 601536 (3)
HOXB2	142967	No OMIM phenotype
HRAS	190020	{Bladder cancer, somatic}, 109800 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant, Isolated cases; Costello syndrome, 218040 (3), Autosomal dominant, Isolated cases; {Nevus sebaceous or woolly hair nevus, somatic}, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); {Spitz nevus or nevus spilus, somatic}, 137550 (3); {Thyroid carcinoma, follicular, somatic}, 188470 (3)
HSPB1	602195	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant
HSPB8	608014	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
HYDIN	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
ID2	600386	No OMIM phenotype
IGF1R	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal recessive, Autosomal dominant
IGFBP4	146733	No OMIM phenotype
IKBKB	603258	Immunodeficiency 15, 615592 (3), Autosomal recessive
ILK	602366	No OMIM phenotype
INHBA	147290	No OMIM phenotype
INHBC	601233	No OMIM phenotype
INPP5D	601582	No OMIM phenotype
INSR	147670	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant; Leprechaunism, 246200 (3), Autosomal recessive; Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive

IRS1	147545	{Coronary artery disease, susceptibility to} (3); {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
IRX4	606199	No OMIM phenotype
ISL1	600366	No OMIM phenotype
ITGAV	193210	No OMIM phenotype
ITGB1	135630	No OMIM phenotype
ITGB3	173470	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant; Glanzmann thrombasthenia, 273800 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3); Purpura, posttransfusion (3); Thrombocytopenia, neonatal alloimmune (3)
ITGB4	147557	Epidermolysis bullosa of hands and feet, 131800 (3), Autosomal dominant; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3), Autosomal recessive
JAG1	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JUN	165160	No OMIM phenotype
KANSL1	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
KAT6A	601408	Mental retardation, autosomal dominant 32, 616268 (3), Autosomal dominant
KAT6B	605880	Genitopatellar syndrome, 606170 (3), Autosomal dominant; SBBYSS syndrome, 603736 (3)
KDM3B	609373	No OMIM phenotype
KDM5A	180202	No OMIM phenotype
KDM5B	605393	No OMIM phenotype
KDM6A	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KDR	191306	Hemangioma, capillary infantile, somatic, 602089 (3); {Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
KIT	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Autosomal dominant, Isolated cases; Germ cell tumors, somatic, 273300 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant; Mast cell disease, 154800 (3), Autosomal dominant; Piebaldism, 172800 (3), Autosomal dominant
KMT2D	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant

KRAS	190070	Bladder cancer, somatic, 109800 (3); Breast cancer, somatic, 114480 (3); Cardiofaciocutaneous syndrome 2, 615278 (3); Gastric cancer, somatic, 137215 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3); Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
KREMEN1	609898	Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive
LAMA2	156225	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive
LAMA5	601033	No OMIM phenotype
LAMC1	150290	No OMIM phenotype
LBR	600024	Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive; Pelger-Huet anomaly, 169400 (3), Autosomal dominant; ?Reynolds syndrome, 613471 (3), Autosomal dominant
LEFTY1	603037	No OMIM phenotype
LEFTY2	601877	Left-right axis malformations (3)
LHX3	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
LMNA	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, AD, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, AR, 616516 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 1B, 159001 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
LMX1B	602575	Nail-patella syndrome, 161200 (3), Autosomal dominant
LRP2	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRRC6	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
LTBP1	150390	No OMIM phenotype
LTBP3	602090	Dental anomalies and short stature, 601216 (3), Autosomal recessive
LZTR1	600574	Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant

MAFG	602020	No OMIM phenotype
MAP2K1	176872	Cardiofaciocutaneous syndrome 3, 615279 (3)
MAP2K2	601263	Cardiofaciocutaneous syndrome 4, 615280 (3)
MAPK14	600289	No OMIM phenotype
MCTP2	616297	No OMIM phenotype
MECOM	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant
MED13L	608771	Mental retardation and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant; Transposition of the great arteries, dextro-looped 1, 608808 (3), Autosomal dominant
MEF2C	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 (3), Autosomal dominant
MEGF8	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MESP1	608689	No OMIM phenotype
MET	164860	?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); {Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3)
METTL3	612472	No OMIM phenotype
MGP	154870	Keutel syndrome, 245150 (3), Autosomal recessive
MID1	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
MITF	156845	COMMAD syndrome, 617306 (3), Autosomal recessive; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; Waardenburg syndrome/ocular albinism, digenic, 103470 (3), Autosomal dominant
MMP2	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
MMP21	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
MSX1	142983	Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3); Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant
MTHFD1	172460	{Abruptio placentae, susceptibility to} (3); {Spina bifida, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTHFR	607093	Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Vascular disease, susceptibility to} (3)

MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MYBPC3	600958	Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
MYC	190080	Burkitt lymphoma, 113970 (3), Isolated cases
MYH10	160776	No OMIM phenotype
MYH11	160745	Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant
MYH6	160710	Atrial septal defect 3, 614089 (3); Cardiomyopathy, dilated, 1EE, 613252 (3); Cardiomyopathy, hypertrophic, 14, 613251 (3), Autosomal dominant; {Sick sinus syndrome 3}, 614090 (3)
MYH7	160760	Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Autosomal dominant; Laing distal myopathy, 160500 (3), Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Scapuloperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant
MYH9	160775	Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant; Epstein syndrome, 153650 (3), Autosomal dominant; Fechtner syndrome, 153640 (3), Autosomal dominant; Macrothrombocytopenia and progressive sensorineural deafness, 600208 (3), Autosomal dominant; May-Hegglin anomaly, 155100 (3), Autosomal dominant; Sebastian syndrome, 605249 (3), Autosomal dominant
MYL2	160781	Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant
MYL3	160790	Cardiomyopathy, hypertrophic, 8, 608751 (3), Autosomal dominant
MYL7	613993	No OMIM phenotype
MYOCD	606127	No OMIM phenotype
MYOM1	603508	No OMIM phenotype
MYOZ2	605602	Cardiomyopathy, hypertrophic, 16, 613838 (3), Autosomal dominant
NF1	613113	Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant
NFATC1	600489	No OMIM phenotype
NFIX	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant
NFKB1	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant

NID2	605399	No OMIM phenotype
NIPBL	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKX2-1	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant
NKX2-5	600584	Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 3, 614432 (3), Autosomal dominant
NKX2-6	611770	Conotruncal heart malformations, 217095 (3); Persistent truncus arteriosus, 217095 (3)
NME8	607421	Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
NNMT	600008	Homocysteine plasma level, 600008 (2)
NODAL	601265	Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant
NONO	300084	Mental retardation, X-linked, syndromic 34, 300967 (3), X-linked
NOS3	163729	{Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant; {Coronary artery spasm 1, susceptibility to} (3); {Hypertension, pregnancy-induced}, 189800 (3), Autosomal dominant; {Hypertension, susceptibility to}, 145500 (3), Multifactorial; {Ischemic stroke, susceptibility to}, 601367 (3), Multifactorial; {Placental abruption} (3)
NOTCH1	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NOTCH2	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NOTCH3	600276	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant; Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant
NPHP3	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive
NPHP4	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive
NPPA	108780	Atrial fibrillation, familial, 6, 612201 (3), Autosomal dominant; Atrial standstill 2, 615745 (3), Autosomal recessive
NR2F2	107773	Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NR4A2	601828	No OMIM phenotype

NRAS	164790	Colorectal cancer, somatic, 114500 (3); Epidermal nevus, somatic, 162900 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Neurocutaneous melanosis, somatic, 249400 (3); Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3)
NRP1	602069	No OMIM phenotype
NRP2	602070	No OMIM phenotype
NSD1	606681	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (1), Autosomal dominant; Sotos syndrome 1, 117550 (3), Autosomal dominant
OTX2	600037	Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant
PACS1	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PAX3	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Autosomal recessive; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant
PCSK5	600488	No OMIM phenotype
PDGFC	608452	No OMIM phenotype
PDGFRA	173490	Gastrointestinal stromal tumor, somatic, 606764 (3); Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3), Isolated cases, Somatic mutation
PDGFRB	173410	Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant
PDX1	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; MODY, type IV, 606392 (3); Pancreatic agenesis 1, 260370 (3), Autosomal recessive
PIGA	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
PIGL	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGN	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive

PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
PITX2	601542	Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Ring dermoid of cornea, 180550 (3), Autosomal dominant
PKD1L1	609721	Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive
PLAGL1	603044	{Diabetes mellitus, transient neonatal}, 601410 (1)
PLAT	173370	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 (1); Thrombophilia, familial, due to decreased release of PLAT, 612348 (1)
POU5F1	164177	No OMIM phenotype
PPARG	601487	Carotid intimal medial thickness 1, 609338 (3); {Diabetes, type 2}, 125853 (3), Autosomal dominant; Insulin resistance, severe, digenic, 604367 (3), Autosomal dominant; Lipodystrophy, familial partial, type 3, 604367 (3), Autosomal dominant; [Obesity, resistance to] (3); Obesity, severe, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
PPARGC1A	604517	No OMIM phenotype
PPP3CA	114105	No OMIM phenotype
PRKACA	601639	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (3)
PRKD1	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
PROX1	601546	No OMIM phenotype
PRRX1	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant
PSEN1	104311	Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant, Isolated cases
PSEN2	600759	Alzheimer disease-4, 606889 (3), Autosomal dominant; Cardiomyopathy, dilated, 1V, 613697 (3), Autosomal dominant

PTEN	601728	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; Malignant melanoma, somatic, 155600 (3); {Meningioma}, 607174 (3), Autosomal dominant; PTEN hamartoma tumor syndrome (3); {Prostate cancer, somatic}, 176807 (3); Squamous cell carcinoma, head and neck, somatic, 275355 (3); VATER association with macrocephaly and ventriculomegaly, 276950 (3), Autosomal recessive
PTK2	600758	No OMIM phenotype
PTP4A3	606449	No OMIM phenotype
PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant
PUF60	604819	Verheij syndrome, 615583 (3), Autosomal dominant
RAB23	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAD21	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant
RAD51C	602774	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive
RAF1	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3)
RAI1	607642	Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases
RAI2	300217	No OMIM phenotype
RARB	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
RARG	180190	No OMIM phenotype
RB1	614041	Bladder cancer, somatic, 109800 (3); Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Autosomal dominant, Somatic mutation; Retinoblastoma, trilateral, 180200 (3), Autosomal dominant, Somatic mutation; Small cell cancer of the lung, somatic, 182280 (3)
RBBP7	300825	No OMIM phenotype
RBL2	180203	No OMIM phenotype
RBM10	300080	TARP syndrome, 311900 (3), X-linked recessive
RBM8A	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
RELA	164014	No OMIM phenotype
RERE	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant

RET	164761	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
RGS19	605071	No OMIM phenotype
RIT1	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RNF20	607699	No OMIM phenotype
ROR2	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RPGR	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3)
RPL11	604175	Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant
RPL15	604174	?Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant
RPL26	603704	?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant
RPL35A	180468	Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant
RPL5	603634	Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant
RPS10	603632	Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant
RPS17	180472	Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant
RPS19	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
RPS24	602412	Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant
RPS26	603701	Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant
RPS28	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant
RPS29	603633	Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant
RPS7	603658	Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant
RSPH1	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
RSPH3	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
RSPH4A	612647	Ciliary dyskinesia, primary, 11, 612649 (3)
RSPH9	612648	Ciliary dyskinesia, primary, 12, 612650 (3)
RYR2	180902	Arrhythmogenic right ventricular dysplasia 2, 600996 (3), Autosomal dominant; Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3), Autosomal dominant
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant; Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant
SALL4	607343	Duane-radial ray syndrome, 607323 (3), Autosomal dominant; IVIC syndrome, 147750 (3), Autosomal dominant

SCN5A	600163	Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; Brugada syndrome 1, 601144 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Long QT syndrome-3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive; Ventricular fibrillation, familial, 1, 603829 (3)
SESN1	606103	No OMIM phenotype
SF3B4	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
SGCA	600119	Muscular dystrophy, limb-girdle, type 2D, 608099 (3), Autosomal recessive
SH3PXD2B	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive
SHH	600725	Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant
SHOC2	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
SKI	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SLC19A1	600424	No OMIM phenotype
SLC25A4	103220	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant
SLC2A10	606145	Arterial tortuosity syndrome, 208050 (3), Autosomal recessive
SLX4	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
SMAD2	601366	No OMIM phenotype
SMAD3	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
SMAD6	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SMARCE1	603111	Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant
SMC1A	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant

SMC3	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SMG9	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive
SOS1	182530	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
SOS2	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOX2	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
SOX4	184430	No OMIM phenotype
SP1	189906	No OMIM phenotype
SPAG1	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
SPATC1L	612412	No OMIM phenotype
SPECC1L	614140	?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant; Opitz GBBB syndrome, type II, 145410 (3), Autosomal dominant
SRCAP	611421	Floating-Harbor syndrome, 136140 (3), Autosomal dominant
SRF	600589	No OMIM phenotype
STAT3	102582	Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant
STRA6	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
SUZ12	606245	No OMIM phenotype
TAB2	605101	Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant
TBX1	602054	Conotruncal anomaly face syndrome, 217095 (3); DiGeorge syndrome, 188400 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TBX2	600747	No OMIM phenotype
TBX20	606061	Atrial septal defect 4, 611363 (3)
TBX3	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
TBX5	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
TBX6	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant
TCAP	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2G, 601954 (3), Autosomal recessive
TCF21	603306	No OMIM phenotype
TCF3	147141	Agammaglobulinemia 8, autosomal dominant, 616941 (3), Autosomal dominant
TCN2	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive

TDGF1	187395	Forebrain defects (3)
TEAD1	189967	Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant
TEK	600221	Glaucoma 3, primary congenital, E, 617272 (3), Autosomal dominant; Venous malformations, multiple cutaneous and mucosal, 600195 (3), Autosomal dominant
TFAP2A	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TFAP2B	601601	Char syndrome, 169100 (3), Autosomal dominant; Patent ductus arteriosus 2, 617035 (3), Autosomal dominant
TFAP2C	601602	No OMIM phenotype
TGFB1	190180	Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive
TGFB2	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant
TGFB3	190230	Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant
TGFBR1	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
TGFBR2	190182	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
TGIF1	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
THBS1	188060	No OMIM phenotype
THBS4	No OMIM gene	No OMIM phenotype
THOC5	612733	No OMIM phenotype
THRB	190160	Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
TKT	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive
TLL1	606742	Atrial septal defect 6, 613087 (3), Autosomal dominant
TNFRSF11A	603499	Osteolysis, familial expansile, 174810 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 7, 612301 (3); {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant
TNFRSF1A	191190	{Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant
TNNC1	191040	Cardiomyopathy, dilated, 1Z, 611879 (3); Cardiomyopathy, hypertrophic, 13, 613243 (3), Autosomal dominant
TNNI3	191044	Cardiomyopathy, dilated, 1FF, 613286 (3); ?Cardiomyopathy, dilated, 2A, 611880 (3), Autosomal recessive; Cardiomyopathy, familial restrictive, 1, 115210 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 7, 613690 (3), Autosomal dominant

TNNT2	191045	Cardiomyopathy, dilated, 1D, 601494 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 3, 612422 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 2, 115195 (3), Autosomal dominant; Left ventricular noncompaction 6, 601494 (3), Autosomal dominant
TNR	601995	No OMIM phenotype
TP53	191170	Adrenal cortical carcinoma, 202300 (3), Autosomal recessive; {Basal cell carcinoma 7}, 614740 (3); Breast cancer, 114480 (3), Autosomal dominant; Choroid plexus papilloma, 260500 (3), Autosomal dominant; Colorectal cancer, 114500 (3), Autosomal dominant; {Glioma susceptibility 1}, 137800 (3), Autosomal dominant, Somatic mutation; Hepatocellular carcinoma, 114550 (3), Somatic mutation; Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Nasopharyngeal carcinoma, 607107 (3); Osteosarcoma, 259500 (3), Autosomal recessive; Pancreatic cancer, 260350 (3), Autosomal dominant, Somatic mutation, Multifactorial
TP73	601990	?Neuroblastoma (1)
TPM1	191010	Cardiomyopathy, dilated, 1Y, 611878 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 3, 115196 (3), Autosomal dominant; Left ventricular noncompaction 9, 611878 (3), Autosomal dominant
TRPM2	603749	No OMIM phenotype
TSR2	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive
TTC25	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
TTN	188840	Cardiomyopathy, dilated, 1G, 604145 (3); Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2J, 608807 (3), Autosomal recessive; Myopathy, proximal, with early respiratory muscle involvement, 603689 (3); Salih myopathy, 611705 (3), Autosomal recessive; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant
TWIST1	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Saethre-Chotzen syndrome with eyelid anomalies, 101400 (3), Autosomal dominant
UBE2B	179095	?Male infertility (1)
UBE2T	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
USP44	610993	No OMIM phenotype
USP9X	300072	Mental retardation, X-linked 99, 300919 (3), X-linked recessive; Mental retardation, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
VCAN	118661	Wagner syndrome 1, 143200 (3), Autosomal dominant

VDR	601769	?Osteoporosis, involutional, 166710 (1), Autosomal dominant; Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
VEGFA	192240	{Microvascular complications of diabetes 1}, 603933 (3)
VEGFC	601528	Lymphedema, hereditary, ID, 615907 (3), Autosomal dominant
WASHC5 (KIAA0196)	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WDR5	No OMIM gene	No OMIM phenotype
WNT11	603699	No OMIM phenotype
WT1	607102	Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Mesothelioma, somatic, 156240 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation
YY1	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
ZEB2	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
ZFHX3	104155	{Prostate cancer, susceptibility to, somatic}, 176807 (3)
ZFPM2	603693	Diaphragmatic hernia 3, 610187 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant; 46XY sex reversal 9, 616067 (3), Autosomal dominant
ZIC3	300265	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
ZMYND10	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: June 06, 2017

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.