CHD

Gene panel

Gene panel information

Gene panel	**CHD**
Version	2
Total genes	233
Activation date	Thursday 03 april 2025
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABCC9	99.92 %	601439	Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive
ABL1	100 %	189980	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation; Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant
ACTA2	99.99 %	102620	Smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant; Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3)
ACTB	100 %	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; Becker nevus, syndromic or isolated, somatic mosaic, 604919 (3); Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 (3), Autosomal dominant; Dystonia-deafness syndrome 1, 607371 (3), Autosomal dominant; Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620479 (3)
ACTC1	98.57 %	102540	Left ventricular noncompaction 4, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant
ACVR1	99.94 %	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ACVR2B	99.99 %	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
ADAM17	99.94 %	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
ADAMTS10	99.99 %	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
ADAMTS19	99.94 %	607513	Cardiac valvular dysplasia 2, 620067 (3), Autosomal recessive
ADNP	100 %	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
AFF4	99.94 %	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
AGO1	99.73 %	606228	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292 (3), Autosomal dominant
ALDH1A2	99.97 %	603687	Diaphragmatic hernia 4, with cardiovascular defects, 620025 (3), Autosomal recessive
AMOTL1	99.96 %	614657	*No OMIM phenotypes*
ANKRD1	99.57 %	609599	*No OMIM phenotypes*
ANKRD11	99.85 %	611192	KBG syndrome, 148050 (3), Autosomal dominant
ANKS3	99.97 %	617310	*No OMIM phenotypes*
ARID1A	99.83 %	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	99.69 %	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARL2BP	99.89 %	615407	Retinitis pigmentosa 82 with or without situs inversus, 615434 (3), Autosomal recessive

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ASXL1	100 %	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
AXIN1	100 %	603816	Hepatocellular carcinoma, somatic, 114550 (3); Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558 (3), Autosomal recessive; ?Caudal duplication anomaly, 607864 (3)
B3GAT3	87.74 %	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
BCL9	99.78 %	602597	*No OMIM phenotypes*
BCOR	99.97 %	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BRAF	99.78 %	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
CACNA1C	100 %	114205	Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3), Autosomal dominant
CAMK2B	99.92 %	607707	Intellectual developmental disorder, autosomal dominant 54, 617799 (3), Autosomal dominant
CASZ1	100 %	609895	*No OMIM phenotypes*
CBL	99.95 %	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CCDC103	99.68 %	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
CCDC22	99.91 %	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
CCDC39	99.74 %	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
CCDC40	100 %	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
CDH2	99.84 %	114020	Arrhythmogenic right ventricular dysplasia 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant
CDK13	99.83 %	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
CFAP53	99.95 %	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
CFC1	21.93 %	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
CHD4	99.99 %	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHRD	99.97 %	603475	*No OMIM phenotypes*
CITED2	100 %	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
COL3A1	99.87 %	120180	Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive
CREBBP	99.97 %	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant

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CRELD1	99.99 %	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; Jeffries-Lakhani neurodevelopmental syndrome, 620771 (3), Autosomal recessive; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
CTNNB1	99.95 %	116806	Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3)
CTNND1	99.92 %	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant
DCHS1	100 %	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
DDX3X	99.01 %	300160	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
DHCR7	99.97 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DLL4	100 %	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DNAAF1	99.99 %	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAAF3	99.99 %	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
DNAAF4	99.78 %	608706	{Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive
DNAH11	99.93 %	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH5	99.98 %	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive
DOCK6	100 %	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DPYSL5	99.93 %	608383	Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant
DVL1	100 %	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
DVL3	99.99 %	601368	Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant
DYRK1A	99.98 %	600855	Intellectual developmental disorder, autosomal dominant 7, 614104 (3), Autosomal dominant
DZIP1	99.97 %	608671	Spermatogenic failure 47, 619102 (3), Autosomal recessive; ?Mitral valve prolapse 3, 610840 (3), Autosomal dominant
EFCAB7	90.45 %	617632	*No OMIM phenotypes*
EFTUD2	99.93 %	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EHMT1	98.38 %	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
ELN	99.86 %	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
EP300	99.97 %	602700	Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
ETS1	99.99 %	164720	*No OMIM phenotypes*
EVC	99.95 %	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	99.97 %	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
FBLN2	99.82 %	135821	*No OMIM phenotypes*

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FBN1	99.85 %	134797	Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant
FBN2	99.9 %	612570	Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant
FGF8	100 %	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGFR2	99.99 %	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant
FLNA	99.99 %	300017	Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
FLT4	97.93 %	136352	Hemangioma, capillary infantile, somatic, 602089 (3); Lymphatic malformation 1, 153100 (3), Autosomal dominant; Congenital heart defects, multiple types, 7, 618780 (3), Autosomal dominant
FOXC1	100 %	601090	Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant
FOXC2	100 %	602402	Lymphedema-distichiasis syndrome, 153400 (3), Autosomal dominant; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3), Autosomal dominant
FOXF1	99.99 %	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FOXH1	100 %	603621	*No OMIM phenotypes*
FOXJ1	100 %	602291	Ciliary dyskinesia, primary, 43, 618699 (3), Autosomal dominant
FOXL1	100 %	603252	Otosclerosis 11, 620576 (3), Autosomal dominant
FOXP1	99.98 %	605515	Intellectual developmental disorder with language impairment with or without autistic features, 613670 (3), Autosomal dominant
GATA4	99.99 %	600576	Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant
GATA5	100 %	611496	Congenital heart defects, multiple types, 5, 617912 (3), Autosomal dominant, Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GATA6	99.9 %	601656	Atrial septal defect 9, 614475 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
GDF1	100 %	602880	Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GJA5	100 %	121013	Atrial fibrillation, familial, 11, 614049 (3), Autosomal dominant; Atrial standstill, digenic (GJA5/SCN5A), 108770 (3), Autosomal dominant
GLI3	100 %	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GPC3	99.6 %	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
GREB1L	99.99 %	617782	Deafness, autosomal dominant 80, 619274 (3), Autosomal dominant; Renal hypodysplasia/aplasia 3, 617805 (3), Autosomal dominant
HAND1	99.99 %	602406	*No OMIM phenotypes*
HAND2	99.97 %	602407	*No OMIM phenotypes*
HAS2	99.99 %	601636	*No OMIM phenotypes*
HDAC8	99.74 %	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HEY2	99.75 %	604674	*No OMIM phenotypes*
HIRA	99.89 %	600237	*No OMIM phenotypes*
HRAS	100 %	190020	Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; 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); Costello syndrome, 218040 (3), Autosomal dominant
INVS	99.94 %	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IRX4	99.99 %	606199	*No OMIM phenotypes*
ISL1	99.99 %	600366	*No OMIM phenotypes*
JAG1	100 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KANSL1	99.85 %	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
KAT6B	99.79 %	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant
KDM5A	99.9 %	180202	El Hayek-Chahrour neurodevelopmental syndrome, 620820 (3), Autosomal recessive
KDM6A	99.74 %	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KDR	99.86 %	191306	{Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3)
KLF13	99.97 %	605328	*No OMIM phenotypes*

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KMT2A	99.97 %	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2D	99.98 %	602113	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant
KRAS	99.13 %	190070	Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)
KYNU	99.19 %	605197	?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive
LEFTY2	100 %	601877	*No OMIM phenotypes*
LRP1	99.94 %	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive; Developmental dysplasia of the hip 3, 620690 (3), Autosomal dominant
LZTR1	99.46 %	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
MAP2K1	99.98 %	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP2K2	99.99 %	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
MATR3	99.86 %	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
MCTP2	99.99 %	616297	*No OMIM phenotypes*
MED12	99.94 %	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
MED13L	99.99 %	608771	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant
MEGF8	99.9 %	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEIS2	99.97 %	601740	Cleft palate, cardiac defects, and impaired intellectual development, 600987 (3), Autosomal dominant
MESP1	100 %	608689	*No OMIM phenotypes*
MKKS	100 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MMP21	99.99 %	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
MRAS	99.97 %	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
MYBPC3	99.98 %	600958	Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
MYH11	99.16 %	160745	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant; Visceral myopathy 2, 619350 (3), Autosomal dominant
MYH6	100 %	160710	{Sick sinus syndrome 3}, 614090 (3); Atrial septal defect 3, 614089 (3); Cardiomyopathy, dilated, 1EE, 613252 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 14, 613251 (3), Autosomal dominant

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MYH7	99.99 %	160760	Laing distal myopathy, 160500 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Congenital myopathy 7B, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Congenital myopathy 7A, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant
MYOM2	99.99 %	603509	*No OMIM phenotypes*
MYRF	99.98 %	608329	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 (3), Autosomal dominant; Cardiac-urogenital syndrome, 618280 (3), Autosomal dominant
NAA15	99.71 %	608000	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant
NDRG4	99.98 %	614463	*No OMIM phenotypes*
NF1	99.88 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
NFATC1	100 %	600489	*No OMIM phenotypes*
NIPBL	99.34 %	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKX2-5	99.75 %	600584	Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant
NKX2-6	100 %	611770	Persistent truncus arteriosus, 217095 (3); Conotruncal heart malformations, 217095 (3)
NODAL	99.98 %	601265	Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant
NONO	99.94 %	300084	Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked
NOTCH1	99.98 %	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NOTCH2	99.03 %	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NPHP3	99.89 %	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NR2F2	100 %	107773	46XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NRAS	99.66 %	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NRP1	99.99 %	602069	*No OMIM phenotypes*
NSD1	99.98 %	606681	Sotos syndrome, 117550 (3), Autosomal dominant
ODAD1	96.04 %	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
OFD1	99.68 %	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
OTUD6B	99.92 %	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
PACS2	99.99 %	610423	Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant
PAN2	99.95 %	617447	*No OMIM phenotypes*
PDGFRA	99.94 %	173490	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 (3); Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3), Isolated cases, Somatic mutation
PIGW	99.87 %	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
PITX2	99.98 %	601542	Ring dermoid of cornea, 180550 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant
PKD1L1	99.84 %	609721	Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive
PLD1	99.82 %	602382	Cardiac valvular dysplasia 1, 212093 (3), Autosomal recessive
PLXND1	99.98 %	604282	Congenital heart defects, multiple types, 9, 620294 (3), Autosomal recessive
POLA1	99.57 %	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
PPP1CB	99.89 %	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PQBP1	99.99 %	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRDM6	99.99 %	616982	Patent ductus arteriosus 3, 617039 (3), Autosomal dominant
PRKD1	99.96 %	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
PTPN11	99.98 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
RAB23	99.97 %	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAF1	99.97 %	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant
RASA2	99.72 %	601589	*No OMIM phenotypes*
RBFOX2	99.99 %	612149	*No OMIM phenotypes*
RERE	99.94 %	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
RIT1	99.78 %	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RLIM	99.93 %	300379	Tonne-Kalscheuer syndrome, 300978 (3), X-linked
RNF40	99.95 %	607700	*No OMIM phenotypes*
ROBO1	99.83 %	602430	Pituitary hormone deficiency, combined or isolated, 8, 620303 (3), Autosomal dominant; Neurooculorenal syndrome, 620305 (3), Autosomal recessive; ?Nystagmus 8, congenital, autosomal recessive, 257400 (3), Autosomal recessive
ROBO4	100 %	607528	Aortic valve disease 3, 618496 (3), Autosomal dominant
ROCK2	99.7 %	604002	*No OMIM phenotypes*
ROR2	99.99 %	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RRAS	99.98 %	165090	*No OMIM phenotypes*
RRAS2	99.94 %	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
SALL1	100 %	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SALL4	100 %	607343	?IVIC syndrome, 147750 (3), Autosomal dominant; Duane-radial ray syndrome, 607323 (3), Autosomal dominant
SF3B4	99.65 %	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
SHOC2	99.96 %	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SHROOM3	99.99 %	604570	*No OMIM phenotypes*
SMAD1	99.96 %	601595	*No OMIM phenotypes*
SMAD2	99.92 %	601366	Loeys-Dietz syndrome 6, 619656 (3), Autosomal dominant; Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657 (3), Autosomal dominant
SMAD3	99.99 %	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
SMAD4	99.97 %	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant
SMAD6	100 %	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SMAD7	99.97 %	602932	{Colorectal cancer, susceptibility to, 3}, 612229 (3)
SMARCA4	99.99 %	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; ?Otosclerosis 12, 620792 (3), Autosomal dominant
SMARCB1	99.99 %	601607	Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant
SMARCC2	99.73 %	601734	Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant
SMARCE1	99.87 %	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
SMC1A	99.98 %	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant
SMC3	99.91 %	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SNAI1	100 %	604238	*No OMIM phenotypes*
SOS1	99.68 %	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SOS2	99.39 %	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOX7	100 %	612202	*No OMIM phenotypes*
SOX9	100 %	608160	Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant; Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant
SPRED2	99.99 %	609292	Noonan syndrome 14, 619745 (3), Autosomal recessive
SRF	99.99 %	600589	*No OMIM phenotypes*
STRA6	99.95 %	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
TAB2	99.75 %	605101	Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant
TBCK	99.75 %	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TBX1	99.95 %	602054	Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TBX18	99.5 %	604613	Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant
TBX20	99.99 %	606061	Atrial septal defect 4, 611363 (3)
TBX3	100 %	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
TBX5	99.98 %	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
TDGF1	99.82 %	187395	*No OMIM phenotypes*
TFAP2B	99.98 %	601601	Patent ductus arteriosus 2, 617035 (3), Autosomal dominant; Char syndrome, 169100 (3), Autosomal dominant
TLL1	99.94 %	606742	Atrial septal defect 6, 613087 (3), Autosomal dominant
TMEM260	99.9 %	617449	Structural heart defects and renal anomalies syndrome, 617478 (3), Autosomal recessive
TRAF7	99.97 %	606692	Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant
UBR1	99.93 %	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
USP34	99.46 %	615295	*No OMIM phenotypes*
USP44	99.86 %	610993	*No OMIM phenotypes*
VEGFA	99.99 %	192240	{Microvascular complications of diabetes 1}, 603933 (3)
VPS35L	99.07 %	618981	Ritscher-Schinzel syndrome 3, 619135 (3), Autosomal recessive
WASHC5	99.98 %	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WDR5	100 %	609012	*No OMIM phenotypes*
WDR62	99.96 %	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
ZEB2	99.97 %	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
ZFPM1	99.82 %	601950	*No OMIM phenotypes*
ZFPM2	100 %	603693	Diaphragmatic hernia 3, 610187 (3); 46XY sex reversal 9, 616067 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
ZFX	94.74 %	314980	Intellectual developmental disorder, X-linked syndromic 37, 301118 (3), X-linked
ZIC3	99.9 %	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

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.