

CardioPathy panel		
versie	V4 (167 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABCC9</i>	601439	Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 10, 608569 (3); Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant
<i>ACADVL</i>	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
<i>ACTA2</i>	102620	Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3); Multisystemic smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant
<i>ACTC1</i>	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
<i>ACTN2</i>	102573	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant
<i>AGL</i>	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
<i>AKAP10</i>	604694	{Cardiac conduction defect, susceptibility to}, 115080 (3), Autosomal dominant
<i>AKAP9</i>	604001	?Long QT syndrome-11, 611820 (3), Autosomal dominant
<i>ALG10</i>	603313	{Long QT syndrome, acquired, reduced susceptibility to}, 613688 (3), Autosomal dominant
<i>ANK2</i>	106410	Cardiac arrhythmia, ankyrin-B-related, 600919 (3), Autosomal dominant; Long QT syndrome 4, 600919 (3), Autosomal dominant
<i>ANKRD1</i>	609599	No OMIM phenotype
<i>ATP5E</i>	606153	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3)
<i>BAG3</i>	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
<i>BRAF</i>	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); Non-small cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
<i>CACNA1C</i>	114205	Brugada syndrome 3, 611875 (3); Timothy syndrome, 601005 (3), Autosomal dominant
<i>CACNA2D1</i>	114204	No OMIM phenotype

<i>CACNB2</i>	600003	Brugada syndrome 4, 611876 (3)
<i>CALM1</i>	114180	Long QT syndrome 14, 616247 (3), Autosomal dominant; Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 (3), Autosomal dominant
<i>CALM2</i>	114182	Long QT syndrome 15, 616249 (3), Autosomal dominant
<i>CALR3</i>	611414	?Cardiomyopathy, hypertrophic, 19, 613875 (3), Autosomal dominant
<i>CASQ2</i>	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive
<i>CAV3</i>	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
<i>CBL</i>	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
<i>CFC1</i>	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
<i>CITED2</i>	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
<i>COA5</i>	613920	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500 (3), Autosomal recessive
<i>CRELD1</i>	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
<i>CRYAB</i>	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
<i>CSRP3</i>	600824	?Cardiomyopathy, dilated, 1M, 607482 (3); Cardiomyopathy, hypertrophic, 12, 612124 (3), Autosomal dominant
<i>CTF1</i>	600435	No OMIM phenotype
<i>CTNNA1</i>	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant
<i>CTNNA3</i>	607667	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616 (3), Autosomal dominant
<i>DCHS1</i>	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive

<i>DES</i>	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; Scapulooperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant
<i>DMD</i>	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
<i>DMPK</i>	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
<i>DNM1L</i>	603850	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal recessive, Autosomal dominant
<i>DOLK</i>	610746	Congenital disorder of glycosylation, type 1m, 610768 (3), Autosomal recessive
<i>DPP6</i>	126141	Mental retardation, autosomal dominant 33, 616311 (3); {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
<i>DSC2</i>	125645	Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant
<i>DSG2</i>	125671	Arrhythmogenic right ventricular dysplasia 10, 610193 (3), Autosomal dominant; Cardiomyopathy, dilated, 1BB, 612877 (3)
<i>DSP</i>	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3); Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive
<i>DTNA</i>	601239	Left ventricular noncompaction 1, with or without congenital heart defects, 604169 (3), Autosomal dominant
<i>ELN</i>	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
<i>EMD</i>	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
<i>EYA4</i>	603550	Cardiomyopathy, dilated, 1J, 605362 (3); Deafness, autosomal dominant 10, 601316 (3), Autosomal dominant
<i>FBN1</i>	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

<i>FBXO32</i>	606604	No OMIM phenotype
<i>FHL1</i>	300163	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant
<i>FHL2</i>	602633	No OMIM phenotype
<i>FKRP</i>	606596	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive
<i>FKTN</i>	607440	Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive
<i>FOXRED1</i>	613622	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>FXN</i>	606829	Friedreich ataxia, 229300 (3), Autosomal recessive; Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive
<i>GAA</i>	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
<i>GATA4</i>	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
<i>GATA5</i>	611496	No OMIM phenotype
<i>GATA6</i>	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
<i>GATAD1</i>	614518	?Cardiomyopathy, dilated, 2B, 614672 (3), Autosomal recessive

<i>GDF1</i>	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
<i>GJA1</i>	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratoderma 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
<i>GJA5</i>	121013	Atrial fibrillation, familial, 11, 614049 (3), Autosomal dominant; Atrial standstill, digenic (<i>GJA5/SCN5A</i>), 108770 (3), Autosomal dominant
<i>GLA</i>	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
<i>GLB1</i>	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
<i>GPD1L</i>	611778	Brugada syndrome 2, 611777 (3)
<i>GUSB</i>	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
<i>HAND1</i>	No OMIM gene	No OMIM phenotype
<i>HCN4</i>	605206	Brugada syndrome 8, 613123 (3); Sick sinus syndrome 2, 163800 (3), Autosomal dominant
<i>HFE</i>	613609	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Hemochromatosis, 235200 (3), Autosomal recessive; {Microvascular complications of diabetes 7}, 612635 (3); {Porphyria cutanea tarda, susceptibility to}, 176100 (3), Autosomal recessive, Autosomal dominant; {Porphyria variegata, susceptibility to}, 176200 (3), Autosomal dominant; [Transferrin serum level QTL2], 614193 (3)
<i>HRAS</i>	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)
<i>ILK</i>	602366	No OMIM phenotype

<i>JAG1</i>	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>JPH2</i>	605267	Cardiomyopathy, hypertrophic, 17, 613873 (3), Autosomal dominant
<i>JUP</i>	173325	Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant; Naxos disease, 601214 (3), Autosomal recessive
<i>KCNA5</i>	176267	Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant
<i>KCND3</i>	605411	Brugada syndrome 9, 616399 (3), Autosomal dominant; Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant
<i>KCNE1</i>	176261	Jervell and Lange-Nielsen syndrome 2, 612347 (3), Autosomal recessive; Long QT syndrome 5, 613695 (3), Autosomal dominant
<i>KCNE2</i>	603796	Atrial fibrillation, familial, 4, 611493 (3); Long QT syndrome 6, 613693 (3), Autosomal dominant
<i>KCNE3</i>	604433	Brugada syndrome 6, 613119 (3)
<i>KCNE5</i>	300328	No OMIM phenotype
<i>KCNH2</i>	152427	Long QT syndrome 2, 613688 (3), Autosomal dominant; {Long QT syndrome 2, acquired, susceptibility to}, 613688 (3), Autosomal dominant; Short QT syndrome 1, 609620 (3)
<i>KCNJ2</i>	600681	Andersen syndrome, 170390 (3), Autosomal dominant; Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3)
<i>KCNJ5</i>	600734	Hyperaldosteronism, familial, type III, 613677 (3), Autosomal dominant; Long QT syndrome 13, 613485 (3), Autosomal dominant
<i>KCNJ8</i>	600935	No OMIM phenotype
<i>KCNQ1</i>	607542	Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Short QT syndrome 2, 609621 (3), Autosomal dominant
<i>KRAS</i>	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)
<i>LAMA4</i>	600133	Cardiomyopathy, dilated, 1J, 615235 (3), Autosomal dominant
<i>LAMP2</i>	309060	Danon disease, 300257 (3), X-linked dominant

<i>LDB3</i>	605906	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Left ventricular noncompaction 3, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant
<i>LMNA</i>	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
<i>MAP2K1</i>	176872	Cardiofaciocutaneous syndrome 3, 615279 (3)
<i>MAP2K2</i>	601263	Cardiofaciocutaneous syndrome 4, 615280 (3)
<i>MED13L</i>	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
<i>MIB1</i>	608677	Left ventricular noncompaction 7, 615092 (3), Autosomal dominant
<i>MRPL3</i>	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
<i>MYBPC3</i>	600958	Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
<i>MYH6</i>	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)
<i>MYH7</i>	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
<i>MYL2</i>	160781	Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant
<i>MYL3</i>	160790	Cardiomyopathy, hypertrophic, 8, 608751 (3), Autosomal dominant

<i>MYLK2</i>	606566	Cardiomyopathy, hypertrophic, 1, digenic, 192600 (3), Autosomal dominant
<i>MYOM1</i>	603508	No OMIM phenotype
<i>MYOZ1</i>	605603	No OMIM phenotype
<i>MYOZ2</i>	605602	Cardiomyopathy, hypertrophic, 16, 613838 (3), Autosomal dominant
<i>MYPN</i>	608517	Cardiomyopathy, dilated, 1KK, 615248 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 4, 615248 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 22, 615248 (3), Autosomal dominant; Nemaline myopathy 11, autosomal recessive, 617336 (3), Autosomal recessive
<i>NEBL</i>	605491	No OMIM phenotype
<i>NEXN</i>	613121	Cardiomyopathy, dilated, 1CC, 613122 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 20, 613876 (3), Autosomal dominant
<i>NKX2-5</i>	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
<i>NKX2-6</i>	611770	Conotruncal heart malformations, 217095 (3); Persistent truncus arteriosus, 217095 (3)
<i>NOS1AP</i>	605551	No OMIM phenotype
<i>NPPA</i>	108780	Atrial fibrillation, familial, 6, 612201 (3), Autosomal dominant; Atrial standstill 2, 615745 (3), Autosomal recessive
<i>NRAS</i>	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)
<i>PDLIM3</i>	605889	No OMIM phenotype
<i>PKP2</i>	602861	Arrhythmogenic right ventricular dysplasia 9, 609040 (3), Autosomal dominant
<i>PLN</i>	172405	Cardiomyopathy, dilated, 1P, 609909 (3); Cardiomyopathy, hypertrophic, 18, 613874 (3), Autosomal dominant
<i>PRKAG2</i>	602743	Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant; Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), ?Autosomal dominant

<i>PSEN1</i>	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
<i>PSEN2</i>	600759	Alzheimer disease-4, 606889 (3), Autosomal dominant; Cardiomyopathy, dilated, 1V, 613697 (3), Autosomal dominant
<i>PTPN11</i>	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
<i>RAF1</i>	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3)
<i>RANGRF</i>	607954	No OMIM phenotype
<i>RBM20</i>	613171	Cardiomyopathy, dilated, 1DD, 613172 (3), Autosomal dominant
<i>RYR2</i>	180902	Arrhythmogenic right ventricular dysplasia 2, 600996 (3), Autosomal dominant; Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3), Autosomal dominant
<i>SCN1B</i>	600235	Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3); Cardiac conduction defect, nonspecific, 612838 (3); Epilepsy, generalized, with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 52, 617350 (3), Autosomal recessive
<i>SCN2B</i>	601327	Atrial fibrillation, familial, 14, 615378 (3), Autosomal dominant
<i>SCN3B</i>	608214	Atrial fibrillation, familial, 16, 613120 (3), Autosomal dominant; Brugada syndrome 7, 613120 (3), Autosomal dominant
<i>SCN4B</i>	608256	Atrial fibrillation, familial, 17, 611819 (3), Autosomal dominant; Long QT syndrome-10, 611819 (3), Autosomal dominant
<i>SCN5A</i>	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)
<i>SCO2</i>	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive; Myopia 6, 608908 (3), Autosomal dominant

<i>SDHA</i>	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant
<i>SGCD</i>	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, type 2F, 601287 (3), Autosomal recessive
<i>SHOC2</i>	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
<i>SLC25A3</i>	600370	Mitochondrial phosphate carrier deficiency, 610773 (3)
<i>SLMAP</i>	602701	No OMIM phenotype
<i>SMAD3</i>	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
<i>SNTA1</i>	601017	Long QT syndrome 12, 612955 (3), Autosomal dominant
<i>SOD2</i>	147460	{Microvascular complications of diabetes 6}, 612634 (3)
<i>SOS1</i>	182530	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
<i>SPRED1</i>	609291	Legius syndrome, 611431 (3), Autosomal dominant
<i>SYNE1</i>	608441	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
<i>SYNE2</i>	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant
<i>TAZ</i>	300394	Barth syndrome, 302060 (3), X-linked recessive
<i>TBX1</i>	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
<i>TBX20</i>	606061	Atrial septal defect 4, 611363 (3)
<i>TCAP</i>	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2G, 601954 (3), Autosomal recessive
<i>TGFB2</i>	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant
<i>TGFB3</i>	190230	Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant
<i>TGFBR1</i>	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
<i>TGFBR2</i>	190182	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
<i>TLL1</i>	606742	Atrial septal defect 6, 613087 (3), Autosomal dominant
<i>TMEM43</i>	612048	Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant

<i>TMEM70</i>	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
<i>TMPO</i>	188380	No OMIM phenotype
<i>TNNC1</i>	191040	Cardiomyopathy, dilated, 1Z, 611879 (3); Cardiomyopathy, hypertrophic, 13, 613243 (3), Autosomal dominant
<i>TNNI3</i>	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
<i>TNNT2</i>	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
<i>TPM1</i>	191010	Cardiomyopathy, dilated, 1Y, 611878 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 3, 115196 (3), Autosomal dominant; Left ventricular noncompaction 9, 611878 (3), Autosomal dominant
<i>TRDN</i>	603283	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441 (3), Autosomal recessive
<i>TRIM63</i>	606131	No OMIM phenotype
<i>TRPM4</i>	606936	Progressive familial heart block, type IB, 604559 (3), Autosomal dominant
<i>TSFM</i>	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
<i>TTN</i>	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
<i>TTR</i>	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
<i>TXNRD2</i>	606448	No OMIM phenotype
<i>VCL</i>	193065	Cardiomyopathy, dilated, 1W, 611407 (3); Cardiomyopathy, hypertrophic, 15, 613255 (3), Autosomal dominant
<i>XK</i>	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
<i>ZFPM2</i>	603693	Diaphragmatic hernia 3, 610187 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant; 46XY sex reversal 9, 616067 (3), Autosomal dominant

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.