CONGENITAL HEART DISEASE GENE PANEL DG 2.16 (58 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACTC1	111,1	100.0%	98.9%	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACVR2B	115,6	99.7%	97.0%	Heterotaxy, visceral, 4, autosomal, 613751
ALDH1A2	105,1	99.9%	98.6%	No OMIM phenotype Tetralogy of Fallot (Pavan (2009) BMC Med Genet 10, 113) Pentalogy of Cantrell (Steiner (2013) J Med Case Rep 7,287) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89,476)
ANKRD1	98,1	99.9%	98.6%	No OMIM phenotype Cardiomyopathy,hypertrophic (Arimura (2009) J Am Coll Cardiol 54,334) Cardiomyopathy,dilated (Duboscq-Bidot (2009) Eur Heart J 30,2128) ?Total anomalous pulmonary venous return (Cinquetti (2008) Hum Mutat 29,468) ?Neurodevelo
BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
CFAP53	131,8	99.1%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	125,8	91.0%	80.1%	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CITED2	149,7	99.2%	99.0%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CRELD1	98,9	99.8%	95.9%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
EHMT1	127,7	94.6%	94.2%	Kleefstra syndrome 1, 610253

ELN	103,1	100.0%	98.9%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
FBN1	137,1	100.0%	99.8%	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FLT4	160,3	99.2%	99.1%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FOXC2	122,3	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXH1	84,5	100.0%	99.5%	No OMIM phenotype Congenital heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J Cardiol 145,83)
FOXL1	144	100.0%	99.4%	No OMIM phenotype ?Hypoplastic left heart syndrome (Iascone (2012) Clin Genet 81,542)
GATA4	87,6	95.9%	86.7%	?Testicular anomalies with or without congenital heart disease, 615542 Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429
GATA5	74	100.0%	99.2%	Congenital heart defects, multiple types, 5, 617912
GATA6	110,2	98.3%	92.5%	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GDF1	50,7	97.8%	84.7%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GJA5	207,8	100.0%	100.0%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
HAND1	162,9	100.0%	100.0%	No OMIM phenotype Ventricular septal defect (Cheng (2011) Clin Chim Acta) Cardiac malformations (Reamon-Buettner (2009) Hum Mol Genet 18,3567) Cardiomyopathy, dilated (Zhou (2015) Clin Chem Lab Med Epub, epub)

HAND2	85,1	100.0%	99.2%	No OMIM phenotype Tetralogy of Fallot (Topf (2014) PLoS One 9,e95453) Ventricular septal defect (Sun (2016) G3 (Bethesda) epub,epub) ?Congenital heart disease (Shen (2010) Chin Med J (Engl) 123,1623)
HEY2	162,7	99.4%	96.6%	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
JAG1	133,7	99.2%	97.1%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920
KRAS	67,2	99.4%	97.3%	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LEFTY2	69,3	99.5%	91.7%	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
MCTP2	120,6	99.5%	97.4%	No OMIM phenotype Coarctation of the aorta (Lalani (2013) Hum Mol Genet 22,4339) ?Bicuspid aortic valve (Bonachea (2014) BMC Med Genomics 7,56)
MED13L	108,5	99.9%	99.6%	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MMP21	94,9	100.0%	98.0%	Heterotaxy, visceral, 7, autosomal, 616749
MYH11	122,1	100.0%	99.5%	Aortic aneurysm, familial thoracic 4, 132900
MYH6	95,5	99.0%	95.3%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	92,2	99.5%	96.4%	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426

				Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
NKX2-5	120,8	100.0%	99.9%	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	139,9	100.0%	100.0%	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NODAL	144,8	100.0%	100.0%	Heterotaxy, visceral, 5, 270100
NOTCH1	141,8	99.8%	98.9%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	123,7	100.0%	99.6%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NR2F2	236,6	100.0%	100.0%	Congenital heart defects, multiple types, 4, 615779
PKD1L1	108,7	100.0%	99.3%	Heterotaxy, visceral, 8, autosomal, 617205
PLD1	116,4	99.9%	99.3%	Cardiac valvular defect, developmental, 212093
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
SHROOM3	151,4	99.9%	99.1%	No OMIM phenotype Heterotaxy (Tariq (2011) Genome Biol 12,R91) ?Neural tube defects (Lemay (2015) J Med Genet 52,493) {Leukaemia risk,association with} (Rudd (2006) Blood 108,638)
SMAD6	180,5	98.8%	89.1%	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
TAB2	170,6	99.9%	99.5%	Congenital heart defects, nonsyndromic, 2, 614980
TBX1	101,2	93.0%	86.9%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400

				Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX20	108,2	100.0%	99.9%	Atrial septal defect 4, 611363
TBX5	135,3	100.0%	100.0%	Holt-Oram syndrome, 142900
TDGF1	120,9	99.7%	94.8%	Forebrain defects, 0
TFAP2B	168,3	99.2%	96.8%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TLL1	129,8	100.0%	99.9%	Atrial septal defect 6, 613087
TNNI3K	105,8	99.9%	99.3%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
ZFPM2	155,6	100.0%	99.8%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	140,9	100.0%	99.8%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors