CONGENITAL HEART DISEASE GENE PANEL DG 2.13 (52 genes)

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACTC1	164.1	100	99	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACVR1	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100
ACVR2B	140.5	97	94	Heterotaxy, visceral, 4, autosomal, 613751
ALDH1A2	114.8	100	99	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	101.7	99	96	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) ?Neurodevelopmental disorder (Handrigan (2013) J Med Genet 50,163)
CFAP53	146.6	97	94	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	74.5	82	71	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	150.7	99	98	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CITED2	111.6	99	99	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CRELD1	114.4	99	97	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRKL	166.4	100	99	No OMIM phenotype ?Congenital heart defect (Breckpot (2012) Am J Med Genet A 158A,574) ?Tetralogy of Fallot (Tomita-Mitchell (2012) Physiol Genomics 44,518) ?Ventricular septal defect (Zhao (2013) Am J Med Genet A 161,3087

ELN	91.1	99	97	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
FBN1	159.8	99	99	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection 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	155.9	98	97	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100
FOXH1	47.2	98	85	No OMIM phenotype Congenital heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J Cardiol 145,83)
GATA4	87.4	68	60	Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429 ?Testicular anomalies with or without congenital heart disease, 615542
GATA5	44.2	98	84	Congenital heart defects, multiple types, 5, 617912
GATA6	61.7	83	72	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	19.5	65	48	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854
GJA5	268.4	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770

HAND1	84.8	100	98	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	32.2	87	67	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	146.1	99	92	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
IRX4	90	95	92	No OMIM phenotype Congenital heart defect (Cheng (2014) BMC Genomics 15,1127) {Prostate cancer,susceptibility to} (Nguyen (2012) Hum Mol Genet 21,2076)
JAG1	148.4	98	97	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
LEFTY2	42.3	91	77	Left-right axis malformations (Koasaki (1999) Am J Hum Genet 64, 712)
MED13L	134.6	100	99	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MMP21	93.3	90	84	Heterotaxy, visceral, 7, autosomal, 616749
MYH11	132.6	100	99	Aortic aneurysm, familial thoracic 4, 132900
MYH6	113.3	99	96	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	111.4	99	96	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Liang distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430

NKX2-5	83.2	100	99	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 Tetrology of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	104.4	100	99	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NODAL	160.7	100	99	Heterotaxy, visceral, 5, 270100
NOTCH1	137.5	99	98	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	172.4	100	99	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NR2F2	246.1	98	94	Congenital heart defects, multiple types, 4, 615779
PITX2	147.8	99	97	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PKD1L1	123.8	100	99	Heterotaxy, visceral, 8, autosomal, 617205
PTPN11	103.1	97	92	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
SHROOM3	137.3	99	98	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	100.5	80	72	Aortic valve disease 2, 614823
TAB2	210.5	99	97	Congenital heart defects, nonsyndromic, 2, 614980
TBX1	75.3	77	67	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX20	142.8	99	99	Atrial septal defect 4, 611363

TBX5	141.3	100	100	Holt-Oram syndrome, 142900
TDGF1	151.4	99	96	Forebrain defects
TFAP2B	153.5	98	96	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TLL1	140.1	100	99	Atrial septal defect 6, 613087
TNNI3K	118.8	98	96	?Cardiac conduction disease with or without dilated cardiomyopathy, 616117
ZFPM2	196.3	100	99	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	113.7	100	99	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 : April 18th, 2018.
This list is accurate for panel version DG 2.13
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors