CONGENITAL HEART DISEASE GENE DG 2.17 (58 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACTC1	118.4	100.0%	99.3%	Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098
ACVR2B	124.2	99.9%	98.3%	Heterotaxy, visceral, 4, autosomal, 613751
ALDH1A2	108.8	99.9%	99.0%	No OMIM Disease ID
ANKRD1	98.9	99.9%	98.6%	No OMIM Disease ID
BRAF	71.0	91.7%	79.4%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
CFAP53	135.5	99.2%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	136.1	91.1%	82.3%	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	143.6	100.0%	99.5%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CITED2	174.9	99.2%	99.2%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CRELD1	105.6	99.9%	96.0%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
EHMT1	138.4	94.7%	94.5%	Kleefstra syndrome 1, 610253
ELN	113.2	100.0%	99.6%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
FBN1	138.3	100.0%	99.7%	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900

FLT4	177.8	99.2%	99.2%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FOXC2	144.2	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXH1	96.2	100.0%	100.0%	No OMIM Disease ID
FOXL1	167.0	100.0%	100.0%	No OMIM Disease ID
GATA4	95.7	98.9%	90.6%	?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429
GATA5	84.0	100.0%	99.5%	Congenital heart defects, multiple types, 5, 617912
GATA6	128.0	99.6%	95.4%	Pancreatic agenesis and congenital heart defects, 600001 Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GDF1	62.1	99.4%	91.2%	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GJA5	225.2	100.0%	100.0%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
HAND1	189.1	100.0%	100.0%	No OMIM Disease ID
HAND2	97.6	100.0%	99.9%	No OMIM Disease ID
HEY2	179.4	99.4%	95.6%	No OMIM Disease ID
JAG1	143.4	99.4%	97.6%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
KMT2D	150.7	100.0%	99.9%	Kabuki syndrome 1, 147920
KRAS	64.0	99.8%	96.8%	Leukemia, acute myeloid, 601626 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215

				Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
LEFTY2	77.2	99.6%	94.4%	No OMIM Disease ID
MCTP2	120.7	99.5%	97.5%	No OMIM Disease ID
MED13L	112.2	100.0%	99.8%	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MMP21	103.7	100.0%	99.5%	Heterotaxy, visceral, 7, autosomal, 616749
MYH11	131.1	100.0%	99.7%	Aortic aneurysm, familial thoracic 4, 132900
MYH6	104.2	99.3%	96.3%	Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 Cardiomyopathy, dilated, 1EE, 613252
MYH7	101.7	99.7%	97.2%	Myopathy, myosin storage, autosomal recessive, 255160 Left ventricular noncompaction 5, 613426 Laing distal myopathy, 160500 Myopathy, myosin storage, autosomal dominant, 608358 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Cardiomyopathy, hypertrophic, 1, 192600
NKX2-5	141.9	100.0%	100.0%	Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	159.7	100.0%	100.0%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NODAL	156.2	100.0%	100.0%	Heterotaxy, visceral, 5, 270100
NOTCH1	158.4	99.8%	99.2%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	130.5	100.0%	99.8%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NR2F2	262.6	100.0%	100.0%	Congenital heart defects, multiple types, 4, 615779
PKD1L1	113.1	100.0%	99.5%	Heterotaxy, visceral, 8, autosomal, 617205
PLD1	116.6	100.0%	99.6%	Cardiac valvular defect, developmental, 212093
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785

RAF1	111.1	100.0%	99.9%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
SHROOM3	165.5	100.0%	99.3%	No OMIM Disease ID
SMAD6	209.5	99.9%	95.6%	Aortic valve disease 2, 614823
SOS1	100.6	99.7%	96.7%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
TAB2	171.5	100.0%	99.7%	Congenital heart defects, nonsyndromic, 2, 614980
TBX1	114.2	93.7%	88.3%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TBX20	112.3	100.0%	100.0%	Atrial septal defect 4, 611363
TBX5	145.6	100.0%	100.0%	Holt-Oram syndrome, 142900
TDGF1	127.1	99.8%	95.5%	Forebrain defects, 0
TFAP2B	186.8	99.7%	97.6%	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TLL1	128.5	100.0%	99.9%	Atrial septal defect 6, 613087
TNNI3K	103.8	100.0%	99.6%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
ZFPM2	160.7	100.0%	99.9%	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZIC3	155.4	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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors