CONGENITAL HEART DISEASE GENE PANEL DG 3.00 (60 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ACTC1	100	99,7	100	100	Left ventricular noncompaction 4, 613424 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098
ACVR2B	98,3	95	100	100	Heterotaxy, visceral, 4, autosomal, 613751
ALDH1A2	99,9	98,5	100	100	No OMIM disease ID
ANKRD1	100	99,4	100	100	No OMIM disease ID
BRAF	91	81,1	100	100	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	99,6	97,4	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	84,2	74,1	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CITED2	99,2	99	100	100	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CRELD1	99,9	95	100	100	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
EHMT1	94,5	93,7	99,6	99,5	Kleefstra syndrome 1, 610253
ELN	99,8	97,8	100	100	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
FBN1	100	99,9	100	100	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	99,2	98,3	100	100	Congenital heart defects, multiple types, 7, 618780 Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FOXC2	100	96,7	100	99,8	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXH1	100	96,5	100	100	No OMIM disease ID
FOXL1	96,6	89	100	100	No OMIM disease ID
GATA4	84,1	74,5	100	99,9	?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	99,7	93,7	100	100	Congenital heart defects, multiple types, 5, 617912
GATA6	89,8	83	99,6	98	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	73,9	54	98,7	92	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJA5	100	100	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770

HAND1	100	100	100	100	No OMIM disease ID
HAND2	99,8	92,6	100	100	No OMIM disease ID
HEY2	100	99,3	100	100	No OMIM disease ID
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 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	88,9	81,4	100	100	No OMIM disease ID
MCTP2	99,7	98,2	100	100	No OMIM disease ID
MED13L	100	99,8	100	100	Transposition of the great arteries, dextro-looped 1, 608808 Mental retardation and distinctive facial features with or without cardiac defects, 616789
MMP21	99,9	98,8	100	100	Heterotaxy, visceral, 7, autosomal, 616749
MYH11	100	100	100	100	Aortic aneurysm, familial thoracic 4, 132900
MYH6	99,4	97,1	100	100	Atrial septal defect 3, 614089 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090 Cardiomyopathy, dilated, 1EE, 613252
MYH7	99,6	97,3	100	100	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
MYRF	99,3	98,5	100	100	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
NKX2-5	100	99,7	100	100	Ventricular septal defect 3, 614432 Tetralogy of Fallot, 187500 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095 Hypothyroidism, congenital nongoitrous, 5, 225250 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	100	99,5	100	100	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NODAL	100	100	100	100	Heterotaxy, visceral, 5, 270100
NOTCH1	99,2	97,2	100	100	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NOTCH2	100	99,5	100	100	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NR2F2	100	98,5	100	100	Congenital heart defects, multiple types, 4, 615779 46,XX sex reversal 5, 618901
PKD1L1	100	99,8	100	100	Heterotaxy, visceral, 8, autosomal, 617205
PLD1	100	99,6	100	100	Cardiac valvular defect, developmental, 212093
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
RAF1	100	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
SHROOM3	98,6	97,8	100	100	No OMIM disease ID
SMAD6	90,9	81	100	99,6	Aortic valve disease 2, 614823 {Radioulnar synostosis, nonsyndromic}, 179300 {Craniosynostosis 7, susceptibility to}, 617439
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
TAB2	100	99,7	100	100	Congenital heart defects, nonsyndromic, 2, 614980

TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TBX20	100	99,7	100	100	Atrial septal defect 4, 611363
TBX5	100	100	100	100	Holt-Oram syndrome, 142900
TDGF1	99,9	96,7	100	100	Forebrain defects, 0
TFAP2B	99,9	98,6	100	100	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TLL1	100	100	100	100	Atrial septal defect 6, 613087
TNNI3K	100	99,4	100	100	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
ZFPM2	100	100	100	100	46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500 Diaphragmatic hernia 3, 610187
ZIC3	100	99,9	100	100	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors