CONGENITAL HEART DISEASE GENE PANEL DG 3.2.0 (65 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ABL1	100	100	100	100	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACTC1	99,9	98,9	100	100	Left ventricular noncompaction 4, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424
ACVR2B	98,6	95,1	100	100	Heterotaxy, visceral, 4, autosomal, 613751
ALDH1A2	100	99	100	100	No OMIM disease ID
ANKRD1	99,9	98,3	100	100	No OMIM disease ID
BRAF	89,4	77,6	100	100	Melanoma, malignant, somatic, 155600 LEOPARD syndrome 3, 613707 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 Noonan syndrome 7, 613706 Colorectal cancer, somatic, 114500 Nonsmall cell lung cancer, somatic, 211980
CFAP53	99,3	96,6	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	85	78	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CHD7	100	99,2	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CITED2	99,2	99,1	100	100	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CRELD1	99,5	94	100	100	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
EHMT1	94,5	93,6	99,6	99,5	Kleefstra syndrome 1, 610253
ELN	99,8	98,3	100	100	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
FBN1	100	99,7	100	100	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328

					Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900
FLT4	99,2	98,9	100	100	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780
FOXC2	100	98,1	100	99,6	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXH1	100	98,7	100	100	No OMIM disease ID
FOXL1	97	88,8	100	100	No OMIM disease ID
GATA4	87,4	78,5	100	100	Tetralogy of Fallot, 187500 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430 ?Testicular anomalies with or without congenital heart disease, 615542
GATA5	99,9	95,5	100	100	Congenital heart defects, multiple types, 5, 617912
GATA6	91,5	84,5	99,7	98,4	Atrial septal defect 9, 614475 Persistent truncus arteriosus, 217095 Pancreatic agenesis and congenital heart defects, 600001 Atrioventricular septal defect 5, 614474 Tetralogy of Fallot, 187500
GDF1	80,8	59	98,5	92	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
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	99,7	94,9	100	100	No OMIM disease ID
HEY2	99,8	98,7	100	100	No OMIM disease ID
JAG1	97,8	96,7	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KDR	100	99,7	100	100	Hemangioma, capillary infantile, somatic, 602089
KMT2D	99,9	99	100	100	Kabuki syndrome 1, 147920
KRAS	99	97,8	100	100	Gastric cancer, somatic, 137215 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
LEFTY2	94,3	84,3	100	100	No OMIM disease ID
MCTP2	99,4	97,7	100	100	No OMIM disease ID
MED13L	100	99,5	100	100	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MMP21	99,8	99,2	100	100	Heterotaxy, visceral, 7, autosomal, 616749
MYH11	100	99,7	100	100	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 Aortic aneurysm, familial thoracic 4, 132900 Visceral myopathy 2, 619350
MYH6	99,2	96,1	100	100	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251
MYH7	99,1	96,7	100	100	Laing distal myopathy, 160500 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430

					Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160
MYRF	99	97,8	100	100	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280
NAA15	94,8	91,2	96,8	96,7	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NKX2-5	100	100	100	100	Hypoplastic left heart syndrome 2, 614435 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Conotruncal heart malformations, variable, 217095 Ventricular septal defect 3, 614432 Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	100	100	100	100	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NODAL	100	100	100	100	Heterotaxy, visceral, 5, 270100
NOTCH1	99,3	97,9	100	100	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	100	99,2	100	100	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NR2F2	100	99,1	100	100	46,XX sex reversal 5, 618901 Congenital heart defects, multiple types, 4, 615779
CCDC114	100	99,8	100	100	Ciliary dyskinesia, primary, 20, 615067
PKD1L1	100	99,3	100	100	Heterotaxy, visceral, 8, autosomal, 617205
PLD1	99,8	98,7	100	100	Cardiac valvular defect, developmental, 212093
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAF1	99,9	99,2	100	100	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
SHROOM3	98,5	97,5	100	100	No OMIM disease ID
SMAD6	90,7	79,3	100	99,9	Aortic valve disease 2, 614823
SOS1	99,6	97,9	100	99,9	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
TAB2	99,8	99,2	100	100	Congenital heart defects, nonsyndromic, 2, 614980

TAF1	99,2	95,7	100	100	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TBX1	87,4	77,6	93,7	90,2	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX20	100	99,8	100	100	Atrial septal defect 4, 611363
TBX5	100	100	100	100	Holt-Oram syndrome, 142900
TDGF1	98,8	91,8	100	100	Forebrain defects,
TFAP2B	98,8	96,4	100	100	Patent ductus arteriosus 2, 617035 Char syndrome, 169100
TLL1	99,9	99,7	100	100	Atrial septal defect 6, 613087
TNNI3K	99,8	99,6	100	100	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
ZFPM2	100	99,9	100	100	Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500
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.
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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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors