CONGENITAL HEART DISEASE GENE PANEL DG 3.5.0 (83 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABL1	100%	100%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACTC1	100%	100%	Left ventricular noncompaction 4, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424
ACVR2B	100%	100%	Heterotaxy, visceral, 4, autosomal, 613751
ADNP	100%	100%	Helsmoortel-van der Aa syndrome, 615873
ALDH1A2	100%	100%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ANKRD1	100%	100%	No OMIM disease ID
ANKRD11	100%	100%	KBG syndrome, 148050
BRAF	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
CACNA1C	100%	100%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 Brugada syndrome 3, 611875
CFAP53	100%	100%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	100%	100%	Heterotaxy, visceral, 2, autosomal, 605376
CHD4	100%	100%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CITED2	100%	100%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431

COL3A1	100%	100%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
CRELD1	100%	100%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
DYRK1A	100%	100%	Intellectual developmental disorder, autosomal dominant 7, 614104
EHMT1	100%	100%	Kleefstra syndrome 1, 610253
ELN	100%	100%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
FBN1	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
FBN2	100%	100%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FLT4	100%	100%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780
FOXH1	100%	100%	No OMIM disease ID
FOXL1	100%	100%	No OMIM disease ID
GATA4	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	100%	100%	Congenital heart defects, multiple types, 5, 617912
GATA6	100%	100%	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	100%	100%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GJA5	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GLYR1	100%	100%	No OMIM disease ID

HAND1	100%	100%	No OMIM disease ID
HAND2	100%	100%	No OMIM disease ID
HEY2	100%	100%	No OMIM disease ID
JAG1	100%	100%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KAT6B	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KDR	100%	100%	Hemangioma, capillary infantile, somatic, 602089
KMT2A	100%	100%	Wiedemann-Steiner syndrome, 605130
KMT2D	100%	100%	Kabuki syndrome 1, 147920
KRAS	100%	100%	Gastric cancer, somatic, 613659 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	100%	100%	No OMIM disease ID
MCTP2	100%	100%	No OMIM disease ID
MED13L	100%	100%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MMP21	100%	100%	Heterotaxy, visceral, 7, autosomal, 616749
MUC16	100%	100%	No OMIM disease ID
MYH11	100%	100%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 Aortic aneurysm, familial thoracic 4, 132900 Visceral myopathy 2, 619350
MYH6	100%	100%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251
MYH7	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	100%	100%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280
NAA15	97%	97%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NF1	100%	100%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NKX2-5	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%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NODAL	100%	100%	Heterotaxy, visceral, 5, 270100
NOTCH1	100%	100%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	100%	100%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NR2F2	100%	100%	46,XX sex reversal 5, 618901 Congenital heart defects, multiple types, 4, 615779
NSD1	100%	100%	Sotos syndrome, 117550
CCDC114	100%	100%	Ciliary dyskinesia, primary, 20, 615067
PKD1L1	100%	100%	Heterotaxy, visceral, 8, autosomal, 617205
PLD1	100%	100%	Cardiac valvular dysplasia 1, 212093
PRKD1	100%	100%	Congenital heart defects and ectodermal dysplasia, 617364
PTPN11	100%	100%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785

RAF1	100%	100%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RBFOX2	100%	100%	No OMIM disease ID
ROBO4	100%	100%	Aortic valve disease 3, 618496
SHROOM3	100%	100%	No OMIM disease ID
SMAD6	100%	100%	Aortic valve disease 2, 614823
SMARCA4	100%	100%	Coffin-Siris syndrome 4, 614609
SOS1	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRF	100%	100%	No OMIM disease ID
TAB2	100%	100%	Congenital heart defects, nonsyndromic, 2, 614980
TAF1	100%	100%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TBX1	98%	96%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX20	100%	100%	Atrial septal defect 4, 611363
TBX5	100%	100%	Holt-Oram syndrome, 142900
TDGF1	100%	100%	Forebrain defects,
TFAP2B	100%	100%	Patent ductus arteriosus 2, 617035 Char syndrome, 169100
TLL1	99%	99%	Atrial septal defect 6, 613087
TMEM260	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TNNI3K	100%	100%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TSC1	100%	100%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
ZFPM2	100%	100%	Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500
ZIC3	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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TWIST X2 is the chemistry used for 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 : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors