CONGENITAL HEARTDISEASE PANEL[1] DG-3.9.0 (95 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABL1	100.0%	100.0%	100.0%	99.4%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACTC1	100.0%	100.0%	100.0%	99.5%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACVR2B	100.0%	100.0%	100.0%	98.7%	Heterotaxy, visceral, 4, autosomal, 613751
ADNP	100.0%	100.0%	100.0%	98.7%	Helsmoortel-van der Aa syndrome, 615873
ALDH1A2	100.0%	99.9%	100.0%	98.4%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ANKRD1	100.0%	99.9%	100.0%	96.2%	
ANKRD11	100.0%	100.0%	100.0%	98.0%	KBG syndrome, 148050

BRAF	100.0%	100.0%	99.9%	96.7%	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	100.0%	100.0%	100.0%	99.0%	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
CCDC114	100.0%	100.0%	100.0%	98.9%	Ciliary dyskinesia, primary, 20, 615067
CFAP45	100.0%	100.0%	100.0%	98.7%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP53	100.0%	100.0%	99.9%	97.2%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	100.0%	100.0%	100.0%	99.6%	Heterotaxy, visceral, 2, autosomal, 605376

CHD4	100.0%	100.0%	100.0%	98.2%	Sifrim-Hitz-Weiss syndrome, 617159
CHD7	100.0%	100.0%	100.0%	98.6%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CITED2	100.0%	100.0%	100.0%	96.5%	Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431
COL3A1	100.0%	100.0%	100.0%	98.1%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
CRELD1	100.0%	100.0%	100.0%	98.7%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217
CTNND1	100.0%	100.0%	100.0%	98.7%	Blepharocheilodontic syndrome 2, 617681
DCHS1	100.0%	100.0%	100.0%	99.7%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DYRK1A	100.0%	100.0%	100.0%	98.7%	Intellectual developmental disorder, autosomal dominant 7, 614104

DZIP1	100.0%	100.0%	100.0%	97.1%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
EHMT1	100.0%	99.9%	99.9%	98.9%	Kleefstra syndrome 1, 610253
ELN	100.0%	100.0%	100.0%	98.8%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
FBN1	100.0%	100.0%	100.0%	99.1%	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	100.0%	100.0%	100.0%	99.4%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

FLNA	100.0%	99.9%	99.0%	78.6%	Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620
FLT4	100.0%	100.0%	100.0%	99.2%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FOXH1	100.0%	100.0%	100.0%	99.2%	
FOXJ1	100.0%	100.0%	100.0%	97.2%	Ciliary dyskinesia, primary, 43, 618699
FOXL1	100.0%	100.0%	99.9%	95.1%	Otosclerosis 11, 620576

GATA4	100.0%	100.0%	99.8%	96.0%	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	100.0%	100.0%	100.0%	97.6%	Congenital heart defects, multiple types, 5, 617912
GATA6	100.0%	100.0%	100.0%	92.6%	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	100.0%	100.0%	100.0%	99.5%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GJA5	100.0%	100.0%	100.0%	99.8%	Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770
GLIS1	100.0%	100.0%	100.0%	99.5%	
GLYR1	100.0%	100.0%	100.0%	98.6%	

HAND1	100.0%	100.0%	100.0%	98.2%	
HAND2	100.0%	100.0%	98.3%	73.0%	
HEY2	100.0%	100.0%	100.0%	98.1%	
JAG1	100.0%	100.0%	100.0%	99.6%	?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	100.0%	100.0%	100.0%	98.4%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KDR	100.0%	100.0%	100.0%	98.5%	{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089
KMT2A	100.0%	100.0%	100.0%	97.9%	Wiedemann-Steiner syndrome, 605130
KMT2D	100.0%	100.0%	100.0%	98.8%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920

KRAS	100.0%	100.0%	100.0%	99.7%	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	100.0%	100.0%	100.0%	99.5%	
LMCD1	100.0%	100.0%	99.9%	98.4%	
MCTP2	100.0%	99.9%	100.0%	98.7%	
MED13L	100.0%	99.6%	100.0%	98.5%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MMP21	100.0%	100.0%	100.0%	98.4%	Heterotaxy, visceral, 7, autosomal, 616749

MUC16	100.0%	100.0%	100.0%	99.3%	
MYH11	100.0%	100.0%	100.0%	98.1%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYH6	100.0%	100.0%	100.0%	97.7%	{Sick sinus syndrome 3}, 614090;Atrial septal defect 3, 614089;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251
MYH7	100.0%	100.0%	100.0%	99.0%	Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358
MYRF	100.0%	100.0%	100.0%	98.6%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280

NAA15	96.6%	96.6%	100.0%	98.3%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NF1	100.0%	100.0%	100.0%	98.6%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NKX2-5	100.0%	100.0%	100.0%	98.2%	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	100.0%	100.0%	100.0%	99.7%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NODAL	100.0%	100.0%	100.0%	99.1%	Heterotaxy, visceral, 5, 270100

NONO	100.0%	99.5%	98.0%	71.3%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOTCH1	100.0%	100.0%	100.0%	99.6%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	100.0%	100.0%	100.0%	99.5%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NR2F2	100.0%	100.0%	99.9%	96.6%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NSD1	100.0%	100.0%	100.0%	98.6%	Sotos syndrome, 117550
PKD1L1	100.0%	100.0%	100.0%	98.8%	Heterotaxy, visceral, 8, autosomal, 617205
PLD1	100.0%	100.0%	100.0%	98.8%	Cardiac valvular dysplasia 1, 212093
PLXND1	100.0%	100.0%	100.0%	98.5%	Congenital heart defects, multiple types, 9, 620294
PPP1R13L	100.0%	99.9%	99.8%	95.7%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519
PRKD1	100.0%	100.0%	99.9%	95.0%	Congenital heart defects and ectodermal dysplasia, 617364

PTPN11	100.0%	100.0%	100.0%	98.2%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
RAF1	100.0%	100.0%	100.0%	98.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RBFOX2	100.0%	100.0%	100.0%	96.5%	
ROBO4	100.0%	100.0%	100.0%	98.9%	Aortic valve disease 3, 618496
SHROOM3	100.0%	100.0%	100.0%	99.5%	
SMAD6	100.0%	100.0%	99.8%	91.5%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
SMARCA4	100.0%	100.0%	100.0%	99.5%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SOS1	100.0%	100.0%	100.0%	96.8%	Noonan syndrome 4, 610733;?Fibromatosis, gingival, 1, 135300
SOX7	100.0%	100.0%	99.8%	92.0%	
SRF	100.0%	100.0%	100.0%	95.7%	

TAB2	100.0%	100.0%	100.0%	98.3%	Congenital heart defects, nonsyndromic, 2, 614980
TAF1	100.0%	99.9%	97.3%	69.0%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TBX1	97.7%	95.5%	99.5%	83.7%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX20	100.0%	100.0%	100.0%	98.5%	Atrial septal defect 4, 611363
TBX5	100.0%	100.0%	100.0%	99.0%	Holt-Oram syndrome, 142900
TDGF1	100.0%	100.0%	100.0%	98.8%	
TFAP2B	100.0%	100.0%	100.0%	98.3%	Patent ductus arteriosus 2, 617035;Char syndrome, 169100
TLL1	99.4%	98.9%	100.0%	98.6%	Atrial septal defect 6, 613087
TMEM260	100.0%	100.0%	100.0%	97.9%	Structural heart defects and renal anomalies syndrome, 617478
TNS1	100.0%	100.0%	100.0%	99.0%	

TSC1	100.0%	100.0%	100.0%	98.8%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
ZFPM2	100.0%	100.0%	100.0%	97.9%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZIC3	100.0%	100.0%	97.4%	68.8%	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.
TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.
TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.
srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : March 17th, 2023.
This list is accurate for panel version DG 3.9.0

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors