CONGENITAL HEARTDISEASE PANEL[1] DG-5.0.0 (130 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
ABL1	100%	100%	100%	100%	99.7%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACTA2	100%	100%	100%	100%	99.8%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042
ACTC1	93.3%	90.8%	100%	100%	99.8%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACVR2B	100%	100%	100%	99.9%	99.5%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS19	94.2%	94%	100%	100%	99.5%	Cardiac valvular dysplasia 2, 620067
ADNP	100%	100%	100%	100%	99.8%	Helsmoortel-van der Aa syndrome, 615873
ALDH1A2	88.9%	88.9%	100%	99.9%	99.5%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ANKRD1	100%	100%	100%	99.8%	99%	
ANKRD11	100%	100%	100%	100%	99.3%	KBG syndrome, 148050
BCL9L	100%	100%	100%	100%	99.5%	

BRAF	100%	100%	100%	99.7%	98.3%	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%	100%	99.9%	99.4%	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
CDK13	100%	100%	100%	100%	99.4%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CFAP45	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP52	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376
CHD4	100%	100%	100%	100%	99.5%	Sifrim-Hitz-Weiss syndrome, 617159

CHD7	100%	100%	100%	100%	99.7%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CIROP	99.7%	96%	100%	100%	99.6%	Heterotaxy, visceral, 12, autosomal, 619702
CIROZ	100%	100%	100%	100%	99.4%	Heterotaxy, visceral, 14, autosomal, 621080
CITED2	100%	100%	100%	100%	100%	Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431
COL3A1	100%	100%	100%	99.9%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
CRELD1	100%	100%	100%	100%	99.3%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPTO	100%	100%	100%	100%	99.9%	
CTNND1	100%	100%	100%	100%	99.6%	Blepharocheilodontic syndrome 2, 617681
DNAH5	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	100%	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 40, 618300
DOCK6	100%	100%	100%	100%	99.3%	Adams-Oliver syndrome 2, 614219
DVL3	100%	100%	100%	100%	99.2%	Robinow syndrome, autosomal dominant 3, 616894

DYRK1A	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 7, 614104
DZIP1	100%	100%	100%	100%	99.4%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
EHMT1	97.7%	97.7%	100%	100%	99.5%	Kleefstra syndrome 1, 610253
EIF3A	100%	100%	100%	100%	99.5%	
EIF3B	100%	100%	100%	100%	99.1%	
ELN	100%	99.9%	100%	99.9%	99.4%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
ETS1	100%	100%	100%	100%	99.7%	
FBN1	100%	100%	100%	100%	99.8%	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	99.2%	99.2%	100%	100%	99.7%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FGF8	100%	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

FLNA	100%	100%	98.4%	87.4%	67%	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	99.7%	98%	100%	99.9%	99.6%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FOXH1	100%	100%	100%	100%	99.7%	
FOXJ1	100%	100%	100%	100%	99%	Ciliary dyskinesia, primary, 43, 618699
GATA4	100%	100%	100%	99.9%	98.2%	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%	99.4%	100%	100%	98.5%	Congenital heart defects, multiple types, 5, 617912

GATA6	93.8%	93.8%	100%	100%	98.6%	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%	100%	100%	100%	99.2%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GJA5	100%	100%	100%	100%	99.8%	Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770
GLIS1	100%	100%	100%	100%	99.2%	
GLYR1	100%	100%	100%	100%	99.4%	
HAND1	100%	100%	100%	100%	98.3%	
HAND2	100%	100%	100%	99.1%	94.6%	
HECTD1	98.5%	98.5%	100%	100%	99.8%	
HEY2	100%	100%	100%	100%	99.3%	
ISL1	100%	100%	100%	100%	99.7%	
JAG1	100%	100%	100%	100%	99.7%	?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%	100%	100%	99.7%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170

KDR	100%	100%	100%	100%	99.8%	{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089
KIF20A	100%	100%	100%	100%	99.7%	?Cardiomyopathy, familial restrictive, 6, 619433
KLF13	100%	100%	100%	99.3%	95.9%	
KMT2A	99.3%	99.2%	100%	100%	99.7%	Wiedemann-Steiner syndrome, 605130
KMT2D	100%	100%	100%	100%	99.1%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KRAS	100%	100%	100%	100%	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

LBX1	100%	100%	100%	99.8%	98.1%	?Central hypoventilation syndrome, congenital, 3, 619483
LMCD1	100%	100%	100%	100%	99.3%	
LZTR1	100%	100%	100%	100%	99.4%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MED13L	100%	100%	100%	99.9%	99.3%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MEGF8	100%	100%	100%	99.8%	98.9%	Carpenter syndrome 2, 614976
MEIS2	89.6%	89.5%	100%	99.9%	99.5%	Cleft palate, cardiac defects, and impaired intellectual development, 600987
MESP1	100%	100%	100%	100%	99.9%	
MMP21	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 7, autosomal, 616749
MNS1	100%	100%	100%	100%	99.9%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MST1R	100%	100%	100%	100%	99.6%	{Nasopharyngeal carcinoma, susceptibility to, 3}, 617075
MUC16	100%	100%	100%	100%	99.5%	
MYH11	100%	100%	100%	100%	99.4%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350

MYH6	100%	100%	100%	100%	99.5%	?Atrial septal defect 3, 614089;{Sick sinus syndrome 3}, 614090;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251
MYH7	99.3%	99.1%	100%	100%	99.5%	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%	99.8%	100%	99.9%	99.1%	Nanophthalmos 1, 600165;Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
NAA15	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NDUFAF1	96.3%	91.9%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NF1	99.4%	99.4%	100%	100%	99.8%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321

NKX2-5	100%	100%	100%	99.9%	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%	100%	100%	100%	99.5%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NODAL	100%	100%	100%	100%	99.5%	Heterotaxy, visceral, 5, autosomal, 270100
NONO	96.7%	90.7%	99.1%	90.1%	68.4%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOTCH1	99.1%	99%	100%	100%	99.2%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	100%	100%	100%	100%	99.7%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NPHP4	100%	100%	100%	99.9%	99.3%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NR2F2	100%	100%	100%	99.9%	98.8%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
ODAD1	100%	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 20, 615067

ODAD2	96.1%	96.1%	100%	100%	99.7%	Ciliary dyskinesia, primary, 23, 615451
OLA1	100%	100%	100%	100%	99.7%	
PITX2	100%	100%	100%	100%	99%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PKD1L1	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 8, autosomal, 617205
PKP2	99.7%	98.4%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLD1	100%	100%	100%	100%	99.6%	Cardiac valvular dysplasia 1, 212093
PLXND1	100%	100%	100%	100%	99.3%	Congenital heart defects, multiple types, 9, 620294
POPDC2	100%	100%	100%	100%	99.8%	Cardiac conduction disease with or without cardiomyopathy 2, 621367
PPP1R13L	100%	100%	100%	99.9%	98.1%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519
PRKD1	100%	100%	100%	99.8%	99.1%	Congenital heart defects and ectodermal dysplasia, 617364
PTPN11	90.5%	89.2%	100%	100%	99.6%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PUF60	100%	100%	100%	100%	99.5%	Verheij syndrome, 615583

RAF1	98.5%	95.2%	100%	100%	99.8%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RBFOX2	85.6%	85.5%	100%	100%	99.5%	
RIT1	100%	100%	100%	100%	99.9%	Noonan syndrome 8, 615355
ROBO4	100%	100%	100%	100%	99.3%	Aortic valve disease 3, 618496
SF3B2	100%	100%	100%	100%	99.5%	Craniofacial microsomia, 164210
SHROOM3	100%	100%	100%	100%	99.6%	
SMAD2	86.7%	86.7%	100%	100%	99.7%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD5	100%	100%	100%	100%	99.9%	
SMAD6	100%	100%	100%	100%	98.6%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
SMARCA4	100%	100%	100%	100%	99.1%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SOS1	98.8%	98.8%	100%	100%	99.8%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOX7	100%	100%	100%	100%	99.2%	
SRF	99.9%	98.1%	100%	100%	99.2%	
STRA6	100%	100%	100%	100%	99.5%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186

TAB2	100%	100%	100%	100%	99.3%	Congenital heart defects, nonsyndromic, 2, 614980
TAF1	98.7%	98.7%	98.5%	87.8%	68.8%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TBX1	98.4%	95.8%	100%	99.9%	97.2%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX20	100%	100%	100%	100%	99.8%	Atrial septal defect 4, 611363
TBX5	100%	100%	100%	100%	99.4%	Holt-Oram syndrome, 142900
TFAP2B	100%	100%	100%	99.9%	98.6%	Patent ductus arteriosus 2, 617035;Char syndrome, 169100
TLL1	100%	100%	100%	100%	99.8%	Atrial septal defect 6, 613087
TMEM260	94.8%	94.8%	100%	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TNS1	100%	100%	100%	100%	99.6%	
TSC1	100%	100%	100%	100%	99.7%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
ZFPM2	100%	100%	100%	100%	99.8%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500

ZIC3	100%	100%	98.6%	84.4%	64.3%	Congenital heart defects, nonsyndromic, multiple types, 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 mapped against GRCh38.
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 mapped against GRCh38.
srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS 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 : November 25th, 2024.
This list is accurate for panel version DG 4.4.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors