CONGENITAL HEART DISEASE GENE PANEL DG 3.3.0 (67 genes)

Releasedate: 13-01-2022

Gene	*TWIST covered >10x*	*TWIST 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
ALDH1A2	100%	100%	No OMIM disease ID
ANKRD1	100%	100%	No OMIM disease ID
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
CFAP53	100%	100%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	100%	100%	Heterotaxy, visceral, 2, autosomal, 605376
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
CRELD1	100%	100%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
EHMT1	99%	99%	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
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%	99%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GJA1	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%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
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
KDR	100%	100%	Hemangioma, capillary infantile, somatic, 602089
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
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	96%	96%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
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
ODAD1	100%	100%	Ciliary dyskinesia, primary, 20, 615067
PKD1L1	100%	100%	Heterotaxy, visceral, 8, autosomal, 617205
PLD1	100%	100%	Cardiac valvular defect, developmental, 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
SHROOM3	100%	100%	No OMIM disease ID
SMAD6	100%	100%	Aortic valve disease 2, 614823
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	97%	94%	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	100%	100%	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
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 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 : January 13th , 2022.
This list is accurate for panel version DG 3.3.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors