# CONGENITAL HEART DISEASE GENE PANEL DG 2.18 (58 genes)

Releasedate: 20-04-2020

| Gene | Agilent V5 covered >10x | Agilent V5 covered > 20x | TWIST covered >10x | TWIST covered >20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|---|
| *ACTC1* | 100% | 99,70% | 100% | 100% | Left ventricular noncompaction 4, 613424<br>Atrial septal defect 5, 612794<br>Cardiomyopathy, dilated, 1R, 613424<br>Cardiomyopathy, hypertrophic, 11, 612098 |
| *ACVR2B* | 98,30% | 95,00% | 100% | 100% | Heterotaxy, visceral, 4, autosomal, 613751 |
| *ALDH1A2* | 99,90% | 98,50% | 100% | 100% | No OMIM disease ID |
| *ANKRD1* | 100% | 99,40% | 100% | 100% | No OMIM disease ID |
| *BRAF* | 95,60% | 85,10% | 100% | 100% | Noonan syndrome 7, 613706<br>Cardiofaciocutaneous syndrome, 115150<br>Adenocarcinoma of lung, somatic, 211980<br>LEOPARD syndrome 3, 613707<br>Nonsmall cell lung cancer, somatic, 0<br>Melanoma, malignant, somatic, 0<br>Colorectal cancer, somatic, 0 |
| *CFAP53* | 99,60% | 97,40% | 100% | 100% | Heterotaxy, visceral, 6, autosomal recessive, 614779 |
| *CFC1* | 84,20% | 74,10% | 100% | 100% | Heterotaxy, visceral, 2, autosomal, 605376 |
| *CHD7* | 100% | 99,50% | 100% | 100% | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370<br>CHARGE syndrome, 214800 |
| *CITED2* | 99,20% | 99,00% | 100% | 100% | Atrial septal defect 8, 614433<br>Ventricular septal defect 2, 614431 |
| *CRELD1* | 99,90% | 95,00% | 100% | 100% | Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 |
| *EHMT1* | 94,50% | 93,70% | 99,60% | 99,50% | Kleefstra syndrome 1, 610253 |
| *ELN* | 99,80% | 97,80% | 100% | 100% | Cutis laxa, autosomal dominant, 123700<br>Supravalvar aortic stenosis, 185500 |
| *FBN1* | 100% | 99,90% | 100% | 100% | Marfan lipodystrophy syndrome, 616914<br>Marfan syndrome, 154700<br>MASS syndrome, 604308<br>Ectopia lentis, familial, 129600<br>Acromicric dysplasia, 102370<br>Weill-Marchesani syndrome 2, dominant, 608328 |

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| | | | | | Geleophysic dysplasia 2, 614185<br>Stiff skin syndrome, 184900 |
| *FLT4* | 99,20% | 98,30% | 100% | 100% | Congenital heart defects, multiple types, 7, 618780<br>Hemangioma, capillary infantile, somatic, 602089<br>Lymphatic malformation 1, 153100 |
| *FOXC2* | 100% | 96,70% | 100% | 99,80% | Lymphedema-distichiasis syndrome, 153400<br>Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 |
| *FOXH1* | 100% | 96,50% | 100% | 100% | No OMIM disease ID |
| *FOXL1* | 96,60% | 89,00% | 100% | 100% | No OMIM disease ID |
| *GATA4* | 84,10% | 74,50% | 100% | 99,90% | ?Testicular anomalies with or without congenital heart disease, 615542<br>Tetralogy of Fallot, 187500<br>Atrioventricular septal defect 4, 614430<br>Atrial septal defect 2, 607941<br>Ventricular septal defect 1, 614429 |
| *GATA5* | 99,70% | 93,70% | 100% | 100% | Congenital heart defects, multiple types, 5, 617912 |
| *GATA6* | 89,80% | 83,00% | 99,60% | 98,00% | Pancreatic agenesis and congenital heart defects, 600001<br>Atrial septal defect 9, 614475<br>Atrioventricular septal defect 5, 614474<br>Persistent truncus arteriosus, 217095<br>Tetralogy of Fallot, 187500 |
| *GDF1* | 73,90% | 54,00% | 98,70% | 92,00% | Right atrial isomerism (Ivemark), 208530<br>Congenital heart defects, multiple types, 6, 613854 |
| *GJA5* | 100% | 100% | 100% | 100% | Atrial fibrillation, familial, 11, 614049<br>Atrial standstill, digenic (GJA5/SCN5A), 108770 |
| *HAND1* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *HAND2* | 99,80% | 92,60% | 100% | 100% | No OMIM disease ID |
| *HEY2* | 100% | 99,30% | 100% | 100% | No OMIM disease ID |
| *JAG1* | 97,70% | 96,80% | 100% | 100% | Alagille syndrome 1, 118450<br>Tetralogy of Fallot, 187500<br>?Deafness, congenital heart defects, and posterior embryotoxon, 617992 |
| *KMT2D* | 100% | 99,40% | 100% | 100% | Kabuki syndrome 1, 147920 |
| *KRAS* | 99,50% | 96,90% | 100% | 100% | Oculoectodermal syndrome, somatic, 600268<br>Leukemia, acute myeloid, somatic, 601626<br>Breast cancer, somatic, 114480<br>RAS-associated autoimmune leukoproliferative disorder, 614470<br>Cardiofaciocutaneous syndrome 2, 615278<br>Arteriovenous malformation of the brain, somatic, 108010<br>Bladder cancer, somatic, 109800 |

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| | | | | | Pancreatic carcinoma, somatic, 260350<br>Lung cancer, somatic, 211980<br>Gastric cancer, somatic, 137215<br>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200<br>Noonan syndrome 3, 609942 |
| *LEFTY2* | 88,90% | 81,40% | 100% | 100% | No OMIM disease ID |
| *MCTP2* | 99,70% | 98,20% | 100% | 100% | No OMIM disease ID |
| *MED13L* | 100% | 99,80% | 100% | 100% | Transposition of the great arteries, dextro-looped 1, 608808<br>Mental retardation and distinctive facial features with or without cardiac defects, 616789 |
| *MMP21* | 99,90% | 98,80% | 100% | 100% | Heterotaxy, visceral, 7, autosomal, 616749 |
| *MYH11* | 100% | 100% | 100% | 100% | Aortic aneurysm, familial thoracic 4, 132900 |
| *MYH6* | 99,40% | 97,10% | 100% | 100% | Atrial septal defect 3, 614089<br>Cardiomyopathy, hypertrophic, 14, 613251<br>Cardiomyopathy, dilated, 1EE, 613252 |
| *MYH7* | 99,60% | 97,30% | 100% | 100% | Myopathy, myosin storage, autosomal recessive, 255160<br>Left ventricular noncompaction 5, 613426<br>Laing distal myopathy, 160500<br>Myopathy, myosin storage, autosomal dominant, 608358<br>Cardiomyopathy, dilated, 1S, 613426<br>Scapuloperoneal syndrome, myopathic type, 181430<br>Cardiomyopathy, hypertrophic, 1, 192600 |
| *NKX2-5* | 100% | 99,70% | 100% | 100% | Ventricular septal defect 3, 614432<br>Hypoplastic left heart syndrome 2, 614435<br>Conotruncal heart malformations, variable, 217095<br>Tetralogy of Fallot, 187500<br>Hypothyroidism, congenital nongoitrous, 5, 225250<br>Atrial septal defect 7, with or without AV conduction defects, 108900 |
| *NKX2-6* | 100% | 99,50% | 100% | 100% | Persistent truncus arteriosus, 217095<br>Conotruncal heart malformations, 217095 |
| *NODAL* | 100% | 100% | 100% | 100% | Heterotaxy, visceral, 5, 270100 |
| *NOTCH1* | 99,20% | 97,20% | 100% | 100% | Aortic valve disease 1, 109730<br>Adams-Oliver syndrome 5, 616028 |
| *NOTCH2* | 100% | 99,50% | 100% | 100% | Hajdu-Cheney syndrome, 102500<br>Alagille syndrome 2, 610205 |
| *NR2F2* | 100% | 98,50% | 100% | 100% | Congenital heart defects, multiple types, 4, 615779 |
| *TAB2* | 100% | 99,70% | 100% | 100% | Congenital heart defects, nonsyndromic, 2, 614980 |
| *PKD1L1* | 100% | 99,80% | 100% | 100% | Heterotaxy, visceral, 8, autosomal, 617205 |
| *PLD1* | 100% | 99,60% | 100% | 100% | Cardiac valvular defect, developmental, 212093 |

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| *PTPN11* | 99,10% | 93,70% | 100% | 100% | LEOPARD syndrome 1, 151100<br>Metachondromatosis, 156250<br>Noonan syndrome 1, 163950<br>Leukemia, juvenile myelomonocytic, somatic, 607785 |
| *RAF1* | 100% | 100% | 100% | 100% | LEOPARD syndrome 2, 611554<br>Noonan syndrome 5, 611553<br>Cardiomyopathy, dilated, 1NN, 615916 |
| *SHROOM3* | 99,90% | 99,10% | 100% | 100% | No OMIM disease ID |
| *SMAD6* | 90,90% | 81,00% | 100% | 99,60% | Aortic valve disease 2, 614823 |
| *SOS1* | 99,80% | 98,40% | 100% | 100% | Noonan syndrome 4, 610733<br>?Fibromatosis, gingival, 1, 135300 |
| *TBX1* | 86,90% | 79,50% | 94,10% | 90,80% | Velocardiofacial syndrome, 192430<br>DiGeorge syndrome, 188400<br>Tetralogy of Fallot, 187500<br>Conotruncal anomaly face syndrome, 217095 |
| *TBX20* | 100% | 99,70% | 100% | 100% | Atrial septal defect 4, 611363 |
| *TBX5* | 100% | 100% | 100% | 100% | Holt-Oram syndrome, 142900 |
| *TDGF1* | 99,90% | 96,70% | 100% | 100% | Forebrain defects, 0 |
| *TFAP2B* | 99,90% | 98,60% | 100% | 100% | Char syndrome, 169100<br>Patent ductus arteriosus 2, 617035 |
| *TLL1* | 100% | 100% | 100% | 100% | Atrial septal defect 6, 613087 |
| *TNNI3K* | 100% | 99,40% | 100% | 100% | Cardiac conduction disease with or without dilated cardiomyopathy, 616117 |
| *ZFPM2* | 100% | 100% | 100% | 100% | 46XY sex reversal 9, 616067<br>Diaphragmatic hernia 3, 610187<br>Tetralogy of Fallot, 187500 |
| *ZIC3* | 100% | 99,90% | 100% | 100% | Congenital heart defects, nonsyndromic, 1, X-linked, 306955<br>Heterotaxy, visceral, 1, X-linked, 306955<br>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.*
*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-DNA coding genes.*
*non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.*
*OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.*

*This list is accurate for panel version DG 2.18*

*Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*