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CARDIAC VALVE ABNORMALITIES PANEL[1] DG-5.1.0 (20 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
ADAMTS10	100%	100%	100%	100%	99.6%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	100%	100%	100%	100%	99.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS19	94%	94%	100%	99.9%	99.3%	Cardiac valvular dysplasia 2, 620067
ADAMTSL2	100%	100%	100%	99.9%	99.4%	Geleophysic dysplasia 1, 231050
AEBP1	100%	100%	100%	99.9%	99.3%	Ehlers-Danlos syndrome, classic-like, 2, 618000
DCHS1	100%	100%	100%	100%	99.6%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DZIP1	100%	100%	100%	100%	99.7%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
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

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FLNA	100%	100%	98.3%	87.4%	68.7%	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
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%	99.1%	100%	99.9%	98.6%	Congenital heart defects, multiple types, 5, 617912
GATA6	93.8%	93.8%	100%	99.8%	98.1%	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
LOX	100%	100%	100%	99.9%	98.5%	Aortic aneurysm, familial thoracic 10, 617168

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LTBP2	100%	100%	100%	100%	99.7%	Glaucoma 3, primary congenital, D, 613086;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;?Weill-Marchesani syndrome 3, recessive, 614819
NKX2-5	100%	100%	100%	99.7%	98.1%	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
NOTCH1	99.1%	99%	100%	99.9%	99.4%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
PLD1	100%	100%	100%	100%	99.8%	Cardiac valvular dysplasia 1, 212093
ROBO4	100%	100%	100%	100%	99.4%	Aortic valve disease 3, 618496
SMAD6	100%	100%	100%	99.8%	98.1%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
TAB2	100%	100%	100%	100%	98.7%	Congenital heart defects, nonsyndromic, 2, 614980

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.

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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 5.0.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors