CARDIAC VALVE ABNORMALITIES PANEL[1] DG-5.2.0 (20 GENES)

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ADAMTS10	100% | 100%	96% | 96%	98% | 98%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	100% | 100%	96% | 96%	99% | 99%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS19	94% | 94%	97% | 97%	99% | 99%	Cardiac valvular dysplasia 2, 620067
ADAMTSL2	99% | 99%	96% | 96%	96% | 94%	Geleophysic dysplasia 1, 231050
AEBP1	100% | 100%	96% | 96%	98% | 98%	Ehlers-Danlos syndrome, classic-like, 2, 618000
DCHS1	100% | 100%	97% | 97%	99% | 99%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DZIP1	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
FBN1	100% | 100%	98% | 98%	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
FLNA	98% | 98%	98% | 98%	99% | 99%	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%	95% | 95%	99% | 99%	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	97% | 97%	94% | 94%	97% | 97%	Congenital heart defects, multiple types, 5, 617912
GATA6	94% | 94%	94% | 94%	99% | 99%	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%	96% | 96%	100% | 100%	Aortic aneurysm, familial thoracic 10, 617168
LTBP2	100% | 100%	97% | 97%	99% | 99%	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%	96% | 96%	99% | 99%	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% | 99%	96% | 96%	98% | 97%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
PLD1	100% | 100%	98% | 98%	100% | 100%	Cardiac valvular dysplasia 1, 212093
ROBO4	100% | 100%	97% | 97%	99% | 99%	Aortic valve disease 3, 618496
SMAD6	100% | 100%	95% | 95%	99% | 99%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
TAB2	100% | 100%	97% | 97%	99% | 99%	Congenital heart defects, nonsyndromic, 2, 614980