

Aortaklep en mitralisklep afwijkingen v3.0 (10 genen, incl. 2core-NL genen)

Gene	% covered > 30x	Associated phenotype description and OMIM ID
DCHS1	>99	Mitral valve prolapse 2, 607829
FLNA	>99	Cardiac valvular dysplasia, X-linked, 314400
GATA4	>99	Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430
GATA5	>99	Congenital heart defects, multiple types, 5
GATA6	>99	Atrial septal defect 9, 614475 Pancreatic agenesis and congenital heart defects, 600001
NKX2.5	>99	Atrial septal defect 7, with or without AV conduction defects, 108900 Hypoplastic left heart syndrome 2, 614435
NOTCH1	>99	Adams-Oliver syndrome 5, 616028 Aortic valve disease, 1109730
ROBO4	>99	Aortic valve disease 8, 618496
SMAD6	>99	Aortic valve disease 2, 614823
TAB2	>99	Congenital heart defects, nonsyndromic, 2, 614980

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014

Genes in bold are core genes

OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015

"No OMIM phenotype" signifies a gene without a current OMIM association

OMIM phenotype descriptions between {} signify risk factors, ? is unconfirmed data