

Supp. Table S1. Cardiac phenotypes of CVM patients

Cardiac phenotype	Patient Group 1*	Non-synonymous variants	Patient Group 2*	Non-synonymous variants	Total
Aortic stenosis	4		19		23
Aortic stenosis with bicuspid aortic valve	4		6	[SMAD6 p.P415L]	10
Atrial septal defect	29		50		79
Atrioventricular septal defect	8		21		29
Bicuspid aortic valve	0		3		3
Coarctation of aorta	3		23		26
Coarctation of aorta with bicuspid aortic valve	4	[SMAD6 p.C484F]#	7		11
Double inlet left ventricle	4		11		15
Ebstein's anomaly	3		6		9
Hypoplastic left heart	2		8		10
Isomerism	2		8		10
Mitral atresia	0		2		2
Mitral valve regurgitation	0		3	[SMAD6: p.A325T]	3
Patent ductus arteriosus	3		15		18
Patent foramen ovale	4		6		10
Pulmonary atresia	0		4		4
Pulmonary atresia + ventricular septal defect	0		10		10
Pulmonary stenosis	2		3		5
Tetralogy of Fallot	0		7		7
Transposition of great arteries	4		52		56
Transposition of great arteries-corrected	0		9		9
Tricuspid atresia	4		12		16
Truncus arteriosus	0		9		9
Ventricular septal defect	10		40		50
Other	0		12		12
Total	90		346		436

Numbers of patient with different cardiac phenotypes in each group are listed.

*The entire coding sequences of *BMP2* (NM_001204.6), *BMP1A* (NM_004329.2) and *SMAD6* (NM_005585.4) genes were sequenced in the first patient group, whilst only the coding region for the MH2 domain of *SMAD6* was sequenced in the second patient group.

This patient also had aortic stenosis.