



Appendix : Cardiovascular Genetics Analysis Panels

Cardiomyopathies	Genes 1 st intent	Genes 2 nd intent	Genes 3 rd intent	
Hypertrophic Cardiomyopathy	LAMP2 MYBPC3 MYH7 PRKAG2 TNNI3	TNNT2 TPM1	ACTC1 ACTN2 ANKRD1 CAV3 CSRP3 GLA JPH2 MYH6 MYL2 MYL3 MYOZ2 NEXN PLN PTPN11 RAF1 TCAP TNNC1 TTR	None
Dilated Cardiomyopathy And Left Ventricular Non-Compaction Cardiomyopathy	BAG3 LDB3 LMNA MYBPC3 MYH6 MYH7	MYPN RBM20 SCN5A TNNI3 TNNT2 TTN	ABCC9 ACTC1 ACTN2 ANKDR1 CSRP3 DES DMD DSC2 DSG2 DSP EMD JUP LAMP2 LAMP2 NEXN PKP2 PLN RAF1 SGCD TAZ TCAP TNNC1 TPM1 VCL	None
Arrhythmogenic Right Ventricular Cardiomyopathy	DSC2 DSG2 DSP	JUP PKP2 TMEM43	DES LMNA PLN RYSR2 SCN5A TGFB3 TTN	None
Amyloidosis	TTR	None	None	
Pan-Cardiomyopathy	Include all genes from HCM, DCM/LVNC, ARVC and Amyloidosis			

Arrhythmias	Genes 1 st intent	Genes 2 nd intent	Genes 3 rd intent
Andersen-Tawil Syndrome	KCNJ2	None	None
Brugada Syndrome	SCN5A	CACNA1C CACNB2 GPD1L KCNE3 SCN1B SCN3B	CACNA2D1 HCN4 KCND3 KCNJ8 PKP2 RANGRF TRPM4
Long QT Syndrome	KCNE1 KCNE2 KCNH2 KCNQ1 SCN5A	AKAP9 ANK2 CACNA1C CALM1 CALM2 CAV3 KCNJ2 KCNJ5 SCN4B SNTA1	None
Short QT Syndrome	KCNH2 KCNJ2 KCNQ1	CACNA1C CACNA2D1 CACNB2	None
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2 KCNJ2 RYSR2	ANK2 CALM1 CALM2 TRDN	None
Unexplained Sudden Cardiac Death	KCNH2 KCNQ1 RYSR2 SCN5A	None	None

Aortopathies	Genes 1 st intent	Genes 2 nd intent	Genes 3 rd intent
Familial Thoracic Aortic Aneurysm	ACTA2	FBN1 SMAD3 TGFB2 TGFB3 TGFB1 TGFB2	MYH11 MYLK SLC2A10
Ehlers-Danlos Syndrome, Type I	COL1A1 COL5A1 COL5A2	COL1A1 only available for specific variant	None
Ehlers-Danlos Syndrome, Type IV	COL3A1	None	None
Loeys-Dietz Syndrome	SMAD3 TGFB2 TGFB3	TGFB1 TGFB2	None
Marfan Syndrome	FBN1	None	None



Familial Dyslipidemia	Genes 1st intent	Genes 2nd intent		Genes 3 rd intent
Familial Hypercholesterolemia	Copy number variation (MLPA) for <i>LDLR</i> gene	<i>APOB</i> <i>APOE</i>	<i>LDLR</i> <i>PCSK9</i>	<i>APOE</i> only if consent form clearly states that patient agrees to be tested
Hyperalphalipoproteinemia	<i>CETP</i>	None		None
Hypertriglyceridemia	<i>LPL</i>	<i>APOA5</i> <i>GPIHBP1</i>	<i>APOC2</i> <i>LMF1</i>	None
Sitosterolemia	<i>ABCG5</i> <i>ABCG8</i>	None		None