

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Apolipoprotein C-II Deficiency	Sanger Sequencing	1	APOC2	267
Arrhythmia	NGS	40	ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CASQ2, CAV3, DES, DSC2, DSG2, DSP, GJA5, GPD1L, HCN4, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ8, KCNQ1, LMNA, NKX2-5, NPPA, PKP2, PLN, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TGFB3, TMEM43	1338
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy	NGS	14	CTNNA3, DES, DSC2, DSG2, DSP, JUP, LDB3, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN	1070
	MLPA	2	DSP, PKP2	590
Brugada Syndrome	NGS	15	CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE5, KCNJ8, RANGRF, SCN5A, SCN1B, SCN2B, SCN3B, TRPM4	1314
	MLPA	1	SCN5A	310
Catecholaminergic Polymorphic Ventricular Tachycardia	NGS	6	ANK2, CALM1, CASQ2, KCNJ2, RYR2, TRDN	1070
Dilated Cardiomyopathy	NGS	44	ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, CRYAB, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, GATAD1, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEXN, PKP2, PLN, RAF1, RBM20, SCN5A, SGCD, TAZ, TBX20, TCAP, TMPO, TNNC1, TNNT2, TNNT2, TPM1, TTN, TTR, VCL	1070
	MLPA	2	BAG3, TNNT2	590
Ehlers-Danlos Syndrome	NGS	37	ADAMTS2, AEBP1, ALDH18A1, ATP7A, ATP6VOA2, B3GALT6, B3GAT3, B4GALT7, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CIR, CIS, GORAB, DSE, EFEMP2, ELN, FBLN5, FBN1, FKBP14, FLNA, LTBP4, PLOD1, PRDM5, PYCR1, SLC39A13, RIN2, SMAD3, TGFB2, TGFB1, TGFB2, TNXB, ZNF469	1070
Familial Hypercholesterolemia	NGS	4	APOB, LDLR, LDLRAP1, PCSK9	1030
	MLPA	1	LDLR	310
Familial Lipoprotein Lipase Deficiency	Sanger Sequencing	1	LPL	267
	MLPA	1	LPL	310
Familial Thoracic Aortic Aneurysm and Dissection and Related Syndromes	NGS	12	ACTA2, COL3A1, COL5A1, FBN1, MYH11, MYLK, SLC2A10, SMAD3, TGFB2, TGFB1, TGFB2, TGFB3	1288
	MLPA	3	FBN1, TGFB1, TGFB2	806
Hyperlipoproteinemia, type 3	Sanger Sequencing	1/2	APOE	87

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Hyperlipoproteinemia, type 5	Sanger Sequencing	1	APOEA5	267
Hyperlipidemia	NGS	6	APOA5, APOC2, GPD1, GPIHBP1, LMF1, LPL	1070
Hypertrophic Cardiomyopathy	NGS	36	ACTC1, ACTN2, AGK, ANKRD1, CALR3, CAV3, CRYAB, CSRP3, FLNC, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, RAF1, SLC25A4, SOS1, TCAP, TNNC1, TNNT2, TPM1, TTN, TTR, VCL	1314
	MLPA	3	MYBPC3, MYH7, TNNT2	806
Lecithin Cholesterol Acyltransferase Deficiency	Sanger Sequencing	1	LCAT	267
Long QT Syndrome	NGS	14	AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN5A, SCN4B, SNTA1	1051
	MLPA	2	KCNH2, KCNQ1	590
Noonan Syndrome	NGS	14	BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1, SPRED1	1051
Pulmonary Arterial Hypertension	NGS	12	ACVRL1, BMPR2, BMPR1B, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, SMAD4, SMAD9	1338
	MLPA	2	ACVRL1, BMPR2	590
Short QT Syndrome	NGS	5	CACNA1C, CACNB2, KCNH2, KCNJ2, KCNQ1	1070
Statin-Induced Myopathy	Sanger Sequencing	1/1	SLCO1B1	87
Tangier Disease	NGS	1	ABCA1	1070
Thrombophilia	RFLP	3/4	F2, F5, MTHFR	87
Whole Exome Sequencing (WES)	NGS		Solo sample	1567
			Trio samples	3100
Whole Genome Sequencing (WGS)	NGS		Solo sample	3635
			Trio samples	7449

Service includes

- DNA extraction
- Genotyping
- Bioinformatic analysis of detected variants
- Detected variants available in Excel format (on request)
- Validation of phenotype/diagnosis associated known or possibly pathogenic variants
- Clinical and biological interpretation
- Results delivery by registered mail (on request)
- TAT 2-9 weeks, excl. WES 10 weeks and WGS 15 weeks

Online ordering, specimen requirements and shipping instructions are available at www.asperbio.com.