

Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Apolipoprotein C-II Deficiency	1	APOC2	Sanger Sequencing	267
Arrhythmia	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	NGS NGS with CNV	1030 1400
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy	14	CTNNA3, DES, DSC2, DSG2, DSP, JUP, LDB3, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN	NGS NGS with CNV	1030 1400
	2	DSP, PKP2	MLPA	590
Brugada Syndrome	15	CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE5, KCNJ8, RANGRF, SCN5A, SCN1B, SCN2B, SCN3B, TRPM4	NGS NGS with CNV	1030 1400
	1	SCN5A	MLPA	310
Catecholaminergic Polymorphic Ventricular Tachycardia	6	ANK2, CALM1, CASQ2, KCNJ2, RYR2, TRDN	NGS NGS with CNV	1030 1400
Dilated Cardiomyopathy	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	NGS NGS with CNV	1030 1400
	2	BAG3, TNNT2	MLPA	590
Ehlers-Danlos Syndrome	37	ADAMTS2, AEBP1, ALDH18A1, ATP7A, ATP6VOA2, B3GALT6, B3GAT3, B4GALT7, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CIR, C1S, GORAB, DSE, EFEMP2, ELN, FBLN5, FBN1, FKBP14, FLNA, LTBP4, PLOD1, PRDM5, PYCR1, SLC39A13, RIN2, SMAD3, TGFB2, TGFB1, TGFB2, TNXB, ZNF469	NGS NGS with CNV	1030 1400
Familial Hypercholesterolemia	4	APOB, LDLR, LDLRAP1, PCSK9	NGS NGS with CNV	1030 1400
	1	LDLR	MLPA	310
Familial Lipoprotein Lipase Deficiency	1	LPL	Sanger Sequencing	267
			MLPA	310
Familial Thoracic Aortic Aneurysm and Dissection and Related Syndromes	12	ACTA2, COL3A1, COL5A1, FBN1, MYH11, MYLK, SLC2A10, SMAD3, TGFB2, TGFB1, TGFB2, TGFB3	NGS NGS with CNV	1030 1400
	3	FBN1, TGFB1, TGFB2	MLPA	806

Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Hereditary Hemorrhagic Telangiectasia	5	ACVRL1, ENG, GDF2, RASA1, SMAD4	NGS NGS with CNV	1030 1400
Hyperlipoproteinemia, type 3	1/2	APOE	Sanger Sequencing	87
Hyperlipoproteinemia, type 5	1	APOEA5	Sanger Sequencing	267
Hypertriglyceridemia	6	APOA5, APOC2, GPD1, GPIHBP1, LMF1, LPL	NGS NGS with CNV	1030 1400
Hypertrophic Cardiomyopathy	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, TNNT3, TPM1, TTN, TTR, VCL	NGS NGS with CNV	1030 1400
	3	MYBPC3, MYH7, TNNT2	MLPA	806
Lecithin Cholesterol Acyltransferase Deficiency	1	LCAT	Sanger Sequencing	267
Long QT Syndrome	14	AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN5A, SCN4B, SNTA1	NGS NGS with CNV	1030 1400
	2	KCNH2, KCNQ1	MLPA	590
Noonan Spectrum Disorders/Rasopathies	25	A2ML1, ACTB, ACTG1, BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1	NGS NGS with CNV	1300 1700
Pulmonary Arterial Hypertension	12	ACVRL1, BMPR2, BMPR1B, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, SMAD4, SMAD9	NGS NGS with CNV	1030 1400
	2	ACVRL1, BMPR2	MLPA	590
Short QT Syndrome	5	CACNA1C, CACNB2, KCNH2, KCNJ2, KCNQ1	NGS NGS with CNV	1030 1400
Statin-Induced Myopathy	1/1	SLCO1B1	Sanger Sequencing	87
Tangier Disease	1	ABCA1	NGS NGS with CNV	1030 1400
Thrombophilia	3/4	F2, F5, MTHFR	RFLP	87
Whole Exome Sequencing (WES)		Solo sample	NGS NGS with CNV	1300 1700
		Trio samples	NGS NGS with CNV	2600 3400
Whole Genome Sequencing (WGS)		Solo sample	NGS	3635
		Trio samples		7449

Version: 2019-06-28

**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](http://www.asperbio.com).