Cardiovascular diseases and RASopathy

Disorder	Gene/Exon (CDS complete)	Quantity/Material	Frequency Test duration	Method
Arrhythmogenic right ventricular cardiomyopathy (ARVC) +	*ANK2, CDH2, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, PLN, PRDM16, RYR2, TMEM43*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Dilated Cardiomyopathy (DCM) +	*ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, DES, DMD, LDB3, DSG2, DSP, EMD, EYA4, FHL1, HFE, LMNA, MYBPC3, MYH6, MYH7, NXN, PLN, PRDM16, PSEN1, PSEN2, RBM20, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNI3, TNNT2, TPM1, TTN, VCL*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Hypertrophic Cardiomyopathy (HCM) +	*ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CALR3, CSRP3, FLNC, GAA, GLA, HRAS, JPH2, KLF10, LAMP2, LZTR1, MAP2K1, MYBC3, MYH6, MYH7, MYL2, MYLK2, MYOM1, MYPN, NRAS, NPC1, NXN, OBSC, PAHX, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SOS1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Left ventricular non-compaction Cardiomyopathy (LVNC) +	*ABCC9, ACTC1, ACTN2, CASQ2, DMPK, DSP, DTNA, HCN4, LDB3, LMNA, MIB1, MYBPC3, MYH7, PKP2, PLEKHM2, PRDM16, RYR2, SCN5A, TAZ, TNNT2, TPM1*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Restrictive Cardiomyopathy (RCM) +	*ABCC6, ACTC1, AGXT, APOA1, BMP5, BMP7, CRYAB, DES, DNAJB6, FHL1, FLNC, GBA, GLA, GRHPR, HAMP, HFE, HFE2, HJV, HOGA1, IDS, IDUA, LBD3, LMNA, MYBPC3, MYH7, MYOT, MYPN, NCP1, NCP2, PNPLA3, SLC40A1, SMPD1, TAZ, TNNC1, TNNI3, TNNT2, TTR, WRN*	5 ml EDTA blood	as needed 4-6 weeks	NGS

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Brugada-Syndrome +	*ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Catecholaminergic polymorphic ventricular tachycardia (CPVT) +	*CALM1, CASQ2, RYR2, TRDN*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Long QT-Syndrome (LQTS) +	*AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Short-QT-Syndrome (SQTS) +	*CACNA1C, CACNB2, KCNH2, KCNJ2, KCNQ1, SCN5A, SLC4A3*	5 ml EDTA blood	as needed 4-6 weeks	NGS
Isolated congenital heart defect +	*ABCC9, ACTC1, ACTA2, ACVR2B, ADAMTS19, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CFAP53, CFC1, CITED2, COL1A1, CREDL1, DNAAF3, DYX1C1, ELN, FBN2, FGFR2, FLNA, GATA4, GATA6, GDF1, GJA1, JAG1, LEFTY2, MMP21, MYH11, MYH6, MYH7, NKX2-5, NODAL, NOTCH1, NOTCH2, NR2F2, SALL4, SOS1, TBX5, ZIC3*	5 ml EDTA blood	as needed 4-6 weeks	NGS

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Disorder	Gene/Exon (CDS complete)	Quantity/Material	Frequency Test duration	Method
Syndromic congenital heart defect +	*ABCC9, ACTB, ADNP, AMER1, ARID1A, ASXL1, BBS2, BBS6, BCOR, BRAF, BRAF1, C14orf104, C19orf51, CBL, CCBE1, CCDC103, CCDC39, CCDC40, CD96, CDK13, CHD4, CHD7, CHST14, COL1A1, COL3A1, CREBBP, CRELD1, DHCR7, DNAH11, DNH5, DNAI1, DNAI2, DNAL1, DYX1C1, EFTUD2, EHMT1, ELN, EP300, EVC, EVC2, FBN2, FGFR2, FLNA, FLNB, FOXC1, FOXC2, FOXF1, GLI3, GPC3, HCCS, HDAC4, HRAS, IFT122, IFT140, INVS, IRX5, JAG1, KANSL1, KDM6A, KMT2A, KMT2D, KRAS, LRRC50, LTBP3, MAP2K1, MAP2K2, MED12, MED13L, MEGF8, MGP, MID1, MKKS, MKS1, NEK1, NEK8, NF1, NFATC1, NIPBL, NME8, NOTCH1, NOTCH2, NPHP3, NRAS, NSD1, PEX1, PEX10, PEX2, PEX26, PEX5, PKD1, PRKD1, PTPN11, RAB23, RAI1, RBM8A, RIT1, SALL1, SALL4, SHOC2, SH3PXD2B, SKI, SLC2A10, SMAD3, SMAD4, SMARCA2, SMARC4, SMARCB1, SMARCE1, SMC1A, SOS1, SOX2, SOX9, STRA6, TAB2, TBX1, TBX3, TBX5, TBX20, TFAP2B, TGFBR1, TGFBR2, UBR1, ZEB2, ZIC3*	5 ml EDTA blood	as needed 4-6 weeks	NGS

Cardiovascular diseases and RASopathy

Disorder	Gene/Exon (CDS complete)	Quantity/Material	Frequency Test duration	Method
RASopathy +	Noonan-Syndrome: *CBL, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, MSH2, MSH6* CFC-Syndrome: *BRAF, KRAS, MAP2K1, MAP2K2* Costello-Syndrome: *HRAS* LEOPARD-Syndrome: *BRAF, MAP2K1, PTPN11, RAF1* Legius-Syndrome: *SPRED1*	5 ml EDTA blood	as needed 4-6 weeks	NGS

Note:
According to the Genetic Diagnostics Act, every examination request for human genetic diagnostics must be accompanied by a declaration of consent from the patient or his legal representative.

Forms for this as well as request forms for the analyses are available in the download area of our homepage.