

Kardiologipanel 2023

Arytmier och kardiomyopatier (93 st)

ABCC9, ACTC1, ACTN2, ALPK3, ALMS1, BAG3, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CNBP, COX15, CRYAB, CSRP3, DES, DMD, DMPK, DSC2, DSG2, DSP, EMD, FHL1, FHOD3, FLNC, FXN, GAA, GJA5, GLA, GNB2, GNB5, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMP2, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, MYL4, MYO6, NEXN, NKX2-5, NPPA, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RANGRF, RBM20, RIT1, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SGO1, SLC25A4, SLMAP, TAFAZZIN, TBX5, TECRL, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL

Arytmier (53 st)

ABCC9, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CNBP, DES, DMD, DMPK, EMD, GJA5, GJC1, GLA, GNB2, GNB5, GPD1L, ABCC9, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CNBP, DES, DMD, DMPK, EMD, GJA5, GJC1, GLA, GNB2, GNB5, GPD1L, HCN4, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMP2, LMNA, MYL4, NKX2-5, NPPA, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SGO1, SLMAP, TBX5, TECRL, TNNI3K, TRDN, TRPM4, TTN

Brugada syndrom, BrS (1 st)

SCN5A

Katekolaminerg polymorf ventrikeltakykardi, CPVT (8 st)

CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN

Förmaksflimmer (11 st)

GJA5, GJC1, KCNA5, KCNH2, KCNQ1, LMNA, MYL4, NPPA, SCN5A, TBX5, TTN

Kort QT-syndrom, SQTS (4 st)

KCNH2, KCNJ2, KCNQ1, SLC4A3

Långt QT-syndrom, LQTS (11 st)

CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN

Kardiell konduktionsstörning, CCD/PCCD (16 st)

CNBP, DES, DMD, DMPK, EMD, GJC1, GLA, LAMP2, LMNA, MYL4, NKX2-5, PRKAG2, SCN5A, TBX5, TNNI3K, TRPM4

Sinusknutedysfunktion, SND (11 st)

CACNA1D, EMD, GNB2, GNB5, HCN4, KCNJ5, KCNQ1, LMNA, RYR2, SCN5A, SGO1

Tidigt repolarisationssyndrom, ERS (19 st)

ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE1L, KCNE3, KCNH2, KCNJ8, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SLMAP, ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE1L, KCNE3, KCNH2, KCNJ8, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SLMAP, TRPM4

Kardiomyopati (54 st)

ABCC9, ACTC1, ACTN2, ALMS1, ALPK3, BAG3, CACNA1C, CAV3, COX15, CRYAB, CSRP3, DES, DSC2, DSG2, DSP, FHL1, FHOD3, FLNC, FXN, GAA, GLA, HCN4, ABCC9, ACTC1, ACTN2, ALMS1, ALPK3, BAG3, CACNA1C, CAV3, COX15, CRYAB, CSRP3, DES, DSC2, DSG2, DSP, FHL1, FHOD3, FLNC, FXN, GAA, GLA, HCN4, JPH2, JUP, LAMP2, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, MYO6, NEXN, NKX2-5, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SLC25A4, TFAZZIN, TBX5, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL

Arytmogen kardiomyopati, ACM (15 st)

BAG3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, NKX2-5, PKP2, PLN, RBM20, SCN5A, TMEM43

Dilaterad kardiomyopati, DCM (19 st)

ACTC1, ACTN2, ALMS1, BAG3, DES, DSP, FLNC, JPH2, LMNA, MYH7, NEXN, PLN, RBM20, SCN5A, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL

Hypertrofisk kardiomyopati, HCM (36 st)

ABCC9, ACTC1, ACTN2, ALPK3, BAG3, CACNA1C, CAV3, COX15, CRYAB, CSRP3, DES, FHL1, FHOD3, FLNC, FXN, GAA, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH7, MYL2, MYL3, MYO6, PLN, PRKAG2, PTPN11, RAF1, RIT1, SLC25A4, TNNC1, TNNI3, TNNT2, TPM1, TTR

Non-compaction kardiomyopati, NCCM (11 st)

ACTC1, HCN4, LDB3, MYBPC3, MYH7, NKX2-5, PRDM16, RYR2, TAZ, TBX5, TTN

Restriktiv kardiomyopati, RCM (7 st)

ACTC1, FLNC, MYH7, TNNI3, TNNT2, TTN, TTR

Kliniskt tillämpbara gener

Kardiovaskulära & metabola gener (27 st)

ACTA2, ACTC1, APOB, CACNA1S, CASQ2, COL3A1, DSC2, DSG2, DSP, DSP, FBN1, FLNC, GAA, GJC1, GLA, HFE, KCNH2, KCNQ1, LDLR, LMNA, MYBPC3, MYH11, MYH7, MYL2, MYL3, PCSK9, PKP2, PRKAG2, RYR2, SCN5A, SMAD3, TGFB1, TGFB2, TMEM43, TNNI3, TNNT2, TPM1, TRDN, TTN

Kardiella gener

ACTC1, CASQ2, DSC2, DSG2, DSP, FLNC, GAA, GLA, KCNH2, KCNQ1, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PRKAG2, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TPM1, TRDN, TTN

Kongenital hjärtsjukdom

Kongenital hjärtsjukdom (141 st)

ABL1, ACTC1, ACVR1, ACVR2B, ADAMTS10, AFF4, ANKRD11, ARID1A, ARID1B, B3GAT3, BCOR, BMP2, BRAF, CDK13, CFC1, CHD4, CHD7, CHST14, CITED2, CREBBP, CRELD1, DLL4, DNAH11, DNAH5, DOCK6, EFTUD2, EHMT1, ELN, EP300, ESCO2, EVC, EVC2, FBN1, FGFR2, FLNA, FLT4, FOXC1, FOXC2, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GLI3, GPC3, HAAO, HAND1, HAND2, HDAC8, HNRNP, HRAS, INVS, JAG1, KANSL1, KAT6A, KAT6B, KDM6A, KDR, KMT2A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAP3K7, MED12, MED13L, MEIS2, MESP1, MYBPC3, MYH11, MYH6, MYH7, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NONO, NOTCH1, NOTCH2, NPHP3, NPHP4, NR2F2, NRAS, NSD1, NUP188, PBX1, PIGL, PIGV, PITX2, PKD1L1, PRDM6, PRKACB, PRKD1, PTPN11, RAB23, RAD21, RAF1, RBFOX2, RERE, RIT1, SALL1, SALL4, SETD5, SF3B4, SHOC2, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMG9, SON, SOS1, STRA6, TAB2, TBX1, TBX20, TBX5, TFAP2B, TGFB1, TGFB2, THOC6, TLL1, TMEM260, TRAF7, TXNL4A, UBR1, WASHC5, ZEB2, ZFPM2, ZIC3

Kongenital hjärtsjukdom, icke syndromal (18 st)

ACVR2B, CCDC11, CFAP53, CFC1, CRELD1, ELN, FLT4, GATA4, NKX2-5, NODAL, NOTCH1, NOTCH1, NR2F2, PKD1L1, TAB2, TBX5, TFAP2B, ZIC3

Referenser:

PMID: 35390533, 33968641, 34012069, 34012068, 34345026, 33324992.
Congenital Heart Disease (CHD): <http://chdgene.victorchang.edu.au/>

Ansvarig:

Maria Mannila, kardiogenetiker