ZENTRUM FÜR LABORMEDIZIN

Molekulargenetik
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Genpanel Kardiopathien

Die zusammengestellen Genlisten basieren auf den aktuellen Empfehlungen der jeweiligen referenzierten Entität. Die Genlisten basierend auf den Panels von Genomics England PanelApp umfassen die als 'diagnostic grade genes' resp. 'high evidence' sowie die 'moderate evidence' Gene.

Der Umfang der Genlisten kann nach Rücksprache mit unserem Labor angepasst und bei Bedarf erweitert resp. reduziert werden.

Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVC) Anzahl Gene: 12 (ARVC_2.1)

ANK2, CDH2, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, PLN, TMEM43

Referenz: Arrhythmogenic right ventricular cardiomyopathy (Version 3.14) green/amber; Genomics England PanelApp

Arrhythmien, kardial Anzahl Gene: 19 (Kardiale Arrhythmien_2.1)

CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, GNB5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, SCN5A, SLC4A3, TANGO2, TECRL, TRDN

Referenz: Cardiac arrhythmias (Version 14.7) green/amber; Genomics England PanelApp

Brugada-Syndrom Anzahl Gene: 2 (Brugada_1.2)

SCN5A, KCNH2

Referenz: Brugada syndrome and cardiac sodium channel disease (Version 3.13) green/amber, Genomics England PanelApp

Dilatative Kardiomyopathie (DCM) Anzahl Gene: 36 (DCM_1.2)

ACTC1, ACTN2, BAG3, CDH2, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, FLNC, JPH2, JUP, LAMP2, LMNA, MYBPC3, MYH7, NEXN, NKX2-5, PKP2, PLN, PPA2, PRDM16, RBM20, RYR2, SCN5A, TBX20, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL

Referenz: Hayesmoore et al., *EJHG*, 2023 EMQN recommendations fpr genetic testing in inherited cardiomyopathies and arrhythmias, PMID: 37443332; Dilated and arrhythmogenic cardiomyopathy (Version 3.1) green; Genomics England PanelApp

Hypertrophe Kardiomyopathie (HCM) Anzahl Gene: 38 (HCM_3.2)

ABCC9, ACTC1, ACTN2, ALPK3, BAG3, CACNA1C, CAV3, COX15, CRYAB, CSRP3, DES, FHL1, FHOD3, FLNC, FXN, GAA, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKAG2, PTPN11, RAF1, RIT1, SLC25A4, TBX20, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTR

Referenz: Arbelo et al., *Eur Heart J*, 2023 ESC Guidelines for the management of cardiomyopathies, PMID: 37622657; Hypertrophic cardiomyopathy (Version 5.1), green; Genomics England PanelApp

Long-QT-Syndrom Anzahl Gene: 12 (LQT_1.1)

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

Referenz: Long QT syndrome (Version 3.11), green/amber; Genomics England PanelApp

Nicht-syndromale kongenitale Herzerkrankungen Anzahl Gene: 29 (NSHK_2.1)

ABL1, ACTC1, ACVR2B, ADAMTS19, CFAP53, CFC1, CRELD1, CTNND1, ELN, FLNA, GATA4, GATA5, GATA6, GDF1, HYAL2, JAG1, LEFTY2, MMP21, MYH6, NKX2-5, NODAL, NOTCH1, NOTCH2, NR2F2, SHROOM3, SPRED2, TAB2, TBX20, TBX5, TLL1, TRAF7, ZIC3

Referenz: Familial non syndromic congenital heart disease (Version 1.87), green/amber; Genomics England PanelApp

Short-QT-Syndrom Anzahl Gene: 8 (SQT_2.1)

CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCN5A, SLC4A3

Referenz: Short QT syndrome (Version 3.15), green/amber, Genomics England PanelApp

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Sudden unexplained death (SUDS) Anzahl Gene: 99 (SUDS_2.1)

ACTC1, ACTN2, ALPK3, ANK2, ANKRD1, ATAD3A, BAG3, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CDH2, CLCA2, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATA6, GLA, GLRA1, GYG1, HCN4, JPH2, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ8, KCNQ1, KLHL24, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYLK3, MYPN, MYZAP, NEXN, NKX2-5, NRAP, PHOX2B, PKP2, PLN, PPA2, PRDM16, PRKAG2, RBM20, RHBDF1, RPL3L, RPS6KB1, RRAGC, RRAGD, RYR2, SCN1B, SCN5A, SGCD, SLC4A3, SLC6A6, SPEG, TAB2, TAX1BP3, TBX20, TBX5, TCAP, TECRL, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSPYL1, TTN, TTR, TULP3, VCL

Referenz: Sudden unexplained death or survivors of a cardiac event (Version 20.6) green/amber & Sudden death in young people (Version 1.15), green/amber, Genomics England PanelApp

Thoraxaorten-Aneurysma, kardiovaskuläre Erkrankungen Anzahl Gene: 34 (FTAAD_2.1)

ABL1, ACTA2, ADAMTSL4, ARIH1, BGN, CBS, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FLCN, FLNA, IPO8, LOX, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2

Referenz: Thoracic aortic aneurysm or dissection (Version 1.127), green/amber; Genomics England PanelApp

Kardiopathien (nicht-syndromal), Gesamtpanel Anzahl Gene: 166 (nicht-synd Kardiopathien_2.1)

ABCC9, ABL1, ACTA2, ACTC1, ACTN2, ACVR2B, ADAMTS19, ADAMTSL4, ALPK3, ANK2, ANKRD1, ARIH1, ATAD3A, BAG3, BGN, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CBS, CDH2, CFAP53, CFC1, CLCA2, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COX15, CRELD1, CRYAB ,CSRP3 ,CTNND1, DES, DMD, DOLK, DSC2, DSG2, DSP, EFEMP2, ELN, EMD, FBLN5, FBN1, FBN2, FHL1, FHOD3, FKRP, FKTN, FLCN, FLII, FLNA, FLNC, FXN, GAA, GATA4, GATA5, GATA6, GDF1, GLA, GLRA1, GNB5, GYG1, HCN4, HYAL2, IPO8, JAG1, JPH2, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ8, KCNQ1, KLHL24, LAMP2, LDB3, LEFTY2, LMNA, LOX, MMP21, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYLK3, MYPN, MYZAP, NEXN, NKX2-5, NODAL, NOTCH1, NOTCH2, NR2F2, NRAP, PHOX2B, PKP2, PLN, PLOD1, PPA2, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RBM20, RHBDF1, RIT1, RPL3L, RPS6KB1, RRAGC, RRAGD, RYR2, SCN1B, SCN5A, SGCD, SHROOM3, SKI, SLC25A4, SLC2A10, SLC4A3, SLC6A6, SMAD2, SMAD3, SMAD4, SPEG, SPRED2, TAB2, TANGO2, TAX1BP3, TBX20, TBX5, TCAP, TECRL, TGFB2, TGFB3, TGFBR1, TGFBR2, TLL1, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRAF7, TRDN, TRIM63, TRPM4, TSPYL1, TTN, TTR, TULP3, VCL, ZIC3

Referenz: ARVC_2.1, Kardiale Arrhythmien_2.1, Brugada_1.2, DCM_1.1, HCM_3.2, LQT_1.1, NSHK_2.1, SQT_2.1, SUDS_2.1, FTAAD_2.1

Syndromale oder pädiatrische Kardiomyopathien Anzahl Gene: 192 (Synd-pädiat Kardiomyopathie_2.1)

AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALMS1, ALPK3, ANK2, ARSB, ATP5D, ATPAF2, BAG3, BRAF, CACNA1C, CAMK2D, CAP2, CASZ1, CBL, CDH2, COA5, COA6, COX10, COX14, COX15, COX20, COX6B1, COX7B, CPT2, CRLS1, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EPG5, EYA4, FAH, FASTKD2, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, FNIP1, FOXRED1, GAA, GATA6, GLA, GLB1, GSN, GUSB, HADHA, HADHB, HCN4, HFE, HGSNAT, HRAS, IDH2, IDS, IDUA, JPH2, JUP, KBTBD13, KRAS, LAMP2, LDB3, LETM1, LMNA, LMOD2, LRPPRC, LZTR1, MAP2K1, MAP2K2, MAP3K7, MIB1, MLYCD, MMACHC, MRAS, MRPL44, MUT, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK3, MYPN, MYZAP, NAA10, NAA15, NAGLU, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA4, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB11, NDUFB3, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEXN, NF1, NKX2-5, NONO, NRAP, NRAS, NUBPL, PCCA, PCCB, PDLIM3, PET100, PKP2, PLD1, PLN, PNPLA2, PPA2, PPCS, PPP1CB, PPP1R13L, PRKAG2, PTPN11, RAF1, RASA2, RBM20, RHBDF1, RIT1, RNF220, RPL3L, RRAGC, RRAGD, RYR2, SCN5A, SCO1, SCO2, SDHA, SDHAF1, SDHD, SGCD, SGSH, SHMT2, SHOC2, SLC22A5, SLC25A20, SLC25A4, SLC30A5, SOS1, SOS2, SPEG, SPRED2, SURF1, TAB2, TAF1A, TAZ, TBX20, TMEM126B, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TSFM, TTN, TTR, UQCC2, VCL

Referenz: Paediatric or syndromic cardiomyopathy (Version 7.1), green/amber; Genomics England PanelApp