

Analyt (Messgröße)	Untersuchungsmaterial	Untersuchungstechnik
Whole genome sequencing (WGS)	EDTA-Blut, DNA	PCR-Free Library Prep, Sequencing by Synthesis
Exome sequencing (ES)	EDTA-Blut, DNA	Sequence Capture, Sequencing-by-synthesis
Virtuelle Panel für WGS und ES für spezifische Diagnosen sind ab Seite 2 dargestellt. Es kann auch eine Auswertung nach HPO-terms oder eine Auswertung eines selbst bestimmten Gen-Panels erfolgen.		
Fluoropyrimidin Toxizität (DPD-Gen: dbSNP rs3918290, rs55886062, rs67376798, rs56038477)	EDTA-Blut, DNA	Genotypisierung mittels LAMP (LaCar)
Morbus Bechterew (HLA-B27 Subtypen B*2702 und B*2705)	EDTA-Blut, DNA	Genotypisierung mittels LAMP (LaCar)
Morbus Bechterew (HLA-B27 Subtypen B*2702, B*2703, B*2704 und B*2705)	EDTA-Blut, DNA	Genotypisierung mittels LAMP (LaCar)
Thrombophilie (F5-Gen: dbSNP rs6025, F2-Gen: rs1799963)	EDTA-Blut, DNA	Genotypisierung mittels LAMP (LaCar)
Hämochromatose (HFE-Gen: rs1800562, rs1799945)	EDTA-Blut, DNA	Genotypisierung mittels LAMP (LaCar)
Siponimod Toxizität (CYP2C9-Gen: CYP2C9*2 (rs1799853) und CYP2C9*3 (rs1057910))	EDTA-Blut, DNA	Genotypisierung mittels LAMP (LaCar)
Mavacamten Toxizität (CYP2C19-Gen: CYP2C19*2, CYP2C19*3 und CYP2C19*17)	EDTA-Blut, DNA	Genotypisierung mittels LAMP (LaCar)
rhAMP-SNP-Genotypisierung zur Detektion von Probenverwechslungen (rs7598901, rs717302, rs354439, rs4688962, rs3124591, rs767809, rs876724, rs2032653)	EDTA-Blut, DNA	qPCR
Segregationsanalysen oder Untersuchung auf Anlageträgerschaften (z.B. <i>ABCC8</i> , <i>BRCA1</i> , <i>BRCA2</i> , <i>BRPF1</i> , <i>DARS2</i> , <i>DICER1</i> , <i>FIG4</i> , <i>MYBPC3</i> , <i>NCDN</i> , <i>NONO</i> , <i>PTPN11</i> , <i>TNNT2</i>)	EDTA-Blut, DNA	PCR und Sanger-Sequenzierung

**Virtuelle Panel für spezifische Verdachtsdiagnosen/Indikationen
im Rahmen von Whole Genome und Exomsequenzierung**

Kardiologische Erkrankungen

<p>Angeborene Herzfehler breit Tier 1: ABL1, ACTC1, CBL, CHD7, ELN, FBN1, FLNA, GATA4, GATA6, GDF1, GJA1, JAG1, KDM6A, KMT2D, KRAS, MAP2K1, MAP2K2, MED13L, NKX2-5, NODAL, NOTCH1, NOTCH2, NR2F2, NRAS, NSD1, PTPN11, RAF1, RIT1, SHOC2, SOS1, TAB2, TBX1, TBX20, TBX5, TGFB1, TGFB2, ZEB2, ARID1A, ASXL1, BCOR, BRAF, CDK13, CFC1, CHD4, CREBBP, EHMT1, EP300, EVC, EVC2, FLT4, HRAS, KDR, LZTR1, MRAS, NF1, PPP1CB, PRKD1, RAI1, RFX2, RRAS2, SALL1, SETBP1, SLC2A10, SOS2, TRAF7</p>
<p>Angeborene Herzfehler breit Tier 2: ACVR2B, CITED2, CRELD1, GATA5, MEIS2, MYH6, NKX2-6, SMAD6, TLL1, ZFP2, ZIC3, ABCC9, ADAMTS10, ADAMTS19, ADNP, AFF4, ALX3, ARHGAP31, ARID1B, ASXL2, ATRX, B3GAT6, B3GAT3, BMP2, CCDC22, CFAP53, CHST3, COL1A2, CSNK2A1, DAND5, DCHS1, DDX11, DLL4, DOCK6, DOHH, DVL3, DYRK1A, EOGT, ESCO2, FBN2, FGFR2, FKBP14, FOXC1, FOXH1, GLA, GPC3, GUSB, HAAO, HAND1, HAND2, HDAC8, HEY2, HNRNPK, HSPA9, HYAL2, HYL1, IDUA, IGFBP7, IPO8, IRX4, KANSL1, KAT6A, KAT6B, KDM5B, KMT2A, KYNU, LAMA4, LEFTY1, LEFTY2, LTBP2, MAP3K7, MAPK1, MAPKAP5, MED12, MEGF8, MID1, MKKS, MMP21, MYH7, MYOM2, NAA10, NADSYN1, NEK1, NIPBL, NONO, NSD2, NSDHL, NUP188, PBX1, PIGL, PKD1L1, PLD1, PLXND1, POLR1A, PRDM6, PRKACA, PRKACB, RAB23, RBM10, RBM8A, RBPJ, RERE, ROBO1, ROBO4, RRAS, SALL4, SF3B4, SH3PXD2B, SHOX2, SKI, SLC37A4, SMAD2, SMAD3, SMAD4, SMC1A, SMC3, SMG9, SMO, SMPD4, SON, SPEN, SPRED2, STK4, STRA6, TBX2, TFAP2B, THOC6, TKT, TMEM260, TMEM94, TXNL4A, UBR7, VANGL2, WASHC5, WBP11, WNT5A, ZMYM2, ZNF699, TTC37</p>
<p>Angeborene Herzfehler nicht syndromal Tier 1: ACTC1, ELN, FBN1, FLNA, GATA4, GATA6, GDF1, GJA1, JAG1, MED13L, NKX2-5, NODAL, NOTCH1, NOTCH2, NR2F2, SOS1, TAB2, TBX1, TBX20, TBX5, TGFB1, TGFB2, CFC1, FLT4, KDR, RFX2, SALL1, SLC2A10</p>
<p>Angeborene Herzfehler nicht syndromal Tier 2: ACVR2B, CITED2, CRELD1, GATA5, MYH6, NKX2-6, SMAD6, TLL1, ZFP2, ZIC3, ABCC9, ADAMTS19, COL1A2, FBN2, FGFR2, FKBP14, FOXH1, HAND1, HAND2, HYAL2, IPO8, IRX4, LAMA4, LEFTY1, LEFTY2, MMP21, MYH7, MYOM2, PBX1, PLXND1, ROBO4, SMAD2, TFAP2B, ABL1, CBL, CHD7, KDM6A, KMT2D, KRAS, MAP2K1, MAP2K2, NRAS, NSD1, PTPN11, RAF1, RIT1, SHOC2, ZEB2, ARID1A, ASXL1, BCOR, BRAF, CDK13, CHD4, CREBBP, EHMT1, EP300, EVC, EVC2, HRAS, LZTR1, MRAS, NF1, PPP1CB, PRKD1, RAI1, RRAS2, SETBP1, SOS2, TRAF7, PRDM6</p>
<p>Arrhythmien Tier 1: ACTC1, ACTN2, ALPK3, BAG3, CACNA1C, CALM1, CALM2, CALM3, CASQ2, CDH2, CSRP3, DES, DMD, DOLK, DSC2, DSP, EMD, FHL1, FHOD3, FLNC, GLA, GLRA1, GNB5, HCN4, JUP, KCNE1, KCNH2, KCNJ2, KCNQ1, LAMP2, LMNA, MYBPC3, MYH7, MYL2, MYL3, NEXN, NKX2-5, PHOX2B, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TSPYL1, TTN, TTR, VCL</p>
<p>Arrhythmien Tier 2: ABCC9, AKAP9, ANK2, ANKRD1, ATAD3A, CACNA2D1, CACNB2, CLCA2, CRYAB, FKRP, FKTN, FLII, GATA6, GPD1L, GYG1, JPH2, KCND3, KCNE2, KCNE3, KCNJ5, KCNJ8, LDB3, MT-TI, MYH6, MYLK2, MYLK3, MYPN, NRAP, PPA2, PRDM16, RHBDF1, RPL3L, RPS6KB1, RRAGC, RRAGD, SCN1B, SCN2B, SCN3B, SCN4B, SGCD, SLC4A3, SLC6A6, SNTA1, SPEG, TAB2, TANGO2, TBX20, TBX5, TCAP, TECL, TRDN, TRPM4, TULP3</p>
<p>ARVC Tier 1: FLNC, DES, DSC2, DSP, JUP, LMNA, TMEM43, PKP2</p>
<p>ARVC Tier 2: ANK2, CDH2, MYH7, PLN, SCN5A, TTN, TGFB3, MYL3, CAV3, CTNNA3, TJP1</p>
<p>DCM Tier 1 : ACTC1, MYH6, NKX2-5, ACTN2, BAG3, CDH2, DES, DMD, DOLK, DSC2, DSP, JUP, LAMP2, LMNA, MYBPC3, MYH7, NEXN, PLN, RBM20, SCN5A, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, EPG5, WWTR1</p>
<p>DCM Tier 2: TBX20, ABCC9, GATA6, TAB2, TBX5, RAF1, ANK2, ANKRD1, CRYAB, FKRP, FKTN, FLII, JPH2, LDB3, MYLK3, MYPN, NRAP, PRDM16, RHBDF1, RPL3L, RRAGC, RRAGD, SCN1B, SGCD, SLC6A6, SPEG, TCAP, CSRP3, FLNC, MYL2, RYR2, TTR, CTF1, DTNA, EYA4, GATAD1, IDH2, ILK, LAMA4, NEBL, PLEKHM2, PSEN1</p>
<p>HCM Tier 1: MYL3, MYH7, PLN, FLNC, ACTC1, ACTN2, CSRP3, LAMP2, MYBPC3, MYL2, TNNC1, TNNI3, TNNT2, TPM1, TTR, GLA, ALPK3, CACNA1C, FHL1, FHOD3, PRKAG2, TRIM63, FXN</p>

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HCM Tier 2: CAV3, ANKRD1, JPH2, MYH6, TCAP, ATAD3A, GYG1, MT-TI, MYLK2, RPS6KB1, TULP3, PDLIM3, TTN, DSP, NEXN, RBM20, RYR2, VCL, CALR3, KLF10, KLHL24, MYOM1, MYOZ2, ABCC9, BAG3, COX15, CRYAB, DES, GAA, LDB3, SLC25A4
Kardiomyopathien Tier 1: ABCC9, ACTC1, ACTN2, BAG3, CDH2, CSRP3, DES, DMD, DOLK, DSC2, DSP, EPG5, FKTN, FLNC, IDH2, MYL3, JUP, LAMP2, PKP2, LMNA, MYBPC3, MYH7, MYL2, NEXN, NKX2-5, PLN, RBM20, SCN5A, WWTR1, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL, GLA, GUSB, IDUA, PLD1, TGFB2, NONO, ALPK3, CACNA1C, CALM1, CALM2, CALM3, CASQ2, EMD, FHL1, FHOD3, GLRA1, HCN4, KCNE1, KCNH2, KCNJ2, KCNQ1, PHOX2B, PRKAG2, TRIM63, TSPYL1, AARS2, ACAD9, ACADVL, ACTA1, AGK, ALMS1, COA5, COA6, COX10, COX15, CPT2, DNAJC19, FXN, GAA, HADHA, HADHB, LMOD2, MLYCD, MRPL44, MMUT, NDUFA11, NDUFA2, NDUFAF1, NDUFB11, NDUFS2, NDUFS8, NDUFV2, PCCA, PCCB, PPCS, PPP1R13L, SCO1, SCO2, SLC22A5, SLC25A20, SLC25A4, TMEM70, TSFM
Kardiomyopathien Tier 2: ANK2, ANKRD1, CAV3, CRYAB, EYA4, FKRP, FLII, GATA6, JPH2, LDB3, MYH6, MYLK3, MYPN, NRAP, PRDM16, TGFB3, RAF1, RHBDF1, RPL3L, RRAGC, RRAGD, RYR2, SCN1B, SGCD, SLC6A6, SPEG, TAB2, TBX20, TBX5, TCAP, BMPR2, NAA10, NAA15, RASA2, RRAS, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2, ATAD3A, CACNA2D1, CACNB2, CLCA2, GYG1, KCND3, KCNE2, KCNJ8, MT-TI, MYLK2, PPA2, RPS6KB1, SLC4A3, TECRL, TRDN, TRPM4, TULP3, AGL, ARSB, ATP5F1D, ATPAF2, CAP2, COX14, COX20, COX6B1, COX7B, CPT1A, CRLS1, ELAC2, FAH, FASTKD2, FNIP1, FOXRED1, GLB1, GSN, HAMP, HFE, HJV, HGSNAT, IDS, LETM1, LRPPRC, MIB1, MMACHC, MTO1, NAGLU, NDUFA1, NDUFA10, NDUFA4, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB3, NDUFB8, NDUFS1, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NUBPL, PDLIM3, PET100, PNPLA2, RNF220, SDHA, SDHAF1, SDHD, SGSH, SHMT2, SLC30A5, SLC40A1, SURF1, TFR2, TMEM126B, TOR1AIP1, UQC2, CTF1, DTNA, GATAD1, ILK, LAMA4, NEBL, PLEKHM2, PSEN1
Long QT Tier 1: SCN5A, KCNQ1, CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNH2, KCNJ2, TRDN
Long QT Tier 2: ANK2, KCNE2, KCNJ5, SCN4B, SNTA1, TECRL
PAH Tier 1: ACVRL1, ATP13A3, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNK3, KDR, SMAD9, SOX17, TBX4
PAH Tier 2: ABCC8, AQP1, BMP10, BMPR1A, BMPR1B, FBLN2, GGCX, KLF2, KLK1, NFU1, NOTCH3, PDGFD, SARS2, SMAD1, SMAD4, TET2
TAAD Tier 1: ABL1, ACTA2, ARIH1, BGN, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FLNA, IPO8, LOX, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFB1, TGFB2
TAAD Tier 2: CBS, FLCN, FOXE3, HCN4, MAT2A, MFAP5, SKI

Sensorineurale Erkrankungen

Sensorineurale Schwerhörigkeit: GJB2

Sensorineurale Schwerhörigkeit Tier 1: ABHD12, ACTG1, ADGRV1, AIFM1, ALMS1, AP1S1, ATP1A3, ATP6V1B1, ATP6V1B2, BCS1L, BMP4, BSND, CABP2, CDC14A, CDC6, CDH23, CDT1, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLDN9, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A5, COL9A1, GSDME, PJVK, DHODH, DIAPH1, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, EIF4A3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FDXR, FGF10, FGF3, FGFR2, FGFR3, FOXI1, FOXI3, FRAS1, FREM2, GATA3, GGPS1, GIPC3, GJB2, GJB3, GJB6, GNAI3, GSPM2, GREB1L, GRHL2, GRIP1, GRXCR1, GSC, GSDME, HAAO, HMX1, HOXA2, HSD17B4, HSPA9, ILDR1, KARS1, KCNE1, KCNJ10, KCNJ16, KCNQ1, KCNQ4, KDM6A, KIT, KMT2D, LARS2, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MASP1, MITF, MN1, MPZL2, MSRB3, MT-RNR1, MT-TS1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OFD1, OGDHL, OPA1, ORC1, ORC4, ORC6, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTX2, P2RX2, PAX2, PAX3, PBX1, PCDH15, PDZD7, PJVK, PLCB4, PLS1, PNPT1, POLR1A, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RNF220, S1PR2, SALL1, SALL4, SERAC1, SERPINB6, SF3B4, SGPL1, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SOX10, SOX2, SPATA5, SPATA5L1, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP, USH1C, USH1G, USH2A, USP48, WFS1, WHRN
Sensorineurale Schwerhörigkeit Tier 2: ABCC1, ACOX1, ATP11A, ATP2B2, BTBD, CACNA1D, CCDC50, CEMIP, CLIC5, CLRN2, COG4, COL4A6, COL9A2, COL9A3, CRLS1, CRYM, DCDC2, DIABLO, DIAPH3, DMXL2, DNAJC3, ELMOD3, ESRP1, FOXF2, GDF6, GRXCR2, HARS2, HGF, HOMER2, KDM3B, KITLG, LETM1, MCM2,

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MET, MIR96, MORC2, NARS2, OXR1, PDSS1, PMP22, PPIP5K2, RIPOR2, ROR1, RPS28, SIX5, SLC26A5, SLC44A4, SNAI2, SPATC1L, SPNS2, SPTBN4, STX4, STXBP3, THOC1, TJP2, TMEM132E, TMTC2, TNC, TOP2B, TSPEAR, TUBB4B, WBP2, YARS1, MAN2B1, ANKH, ARSG, CDKN1C, EXOSC2, IARS2, MANBA, PEX1, PEX6, PEX26, PHYH, PSIP1, SGO2, TWNK, TYR

Retinale Erkrankungen Tier 1: ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, AFG3L2, AGBL5, AH11, AIPL1, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARSG, ATF6, ATOH7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CFAP410, PCARE, CFAP418, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP250, CEP290, CEP78, CERKL, CFAP20, CFH, CHM, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL18A1, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COQ2, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTNND1, CTSD, CWC27, CYP4V2, DHDDS, DRAM2, DYNC2H1, EFEMP1, ELOVL4, ERCC6, ERCC8, EYS, FAM161A, TLCD3B, FLVCR1, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HCCS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT74, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIAA1549, KIF11, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MCOLN1, MED12, MERTK, MFRP, MFSD8, MIR204, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MTPP, MVK, MYO7A, NBAS, NDP, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPN1LW, OPN1MW, OTX2, P3H2, PANK2, PAX2, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDSS1, PEX1, PEX2, PEX6, PEX7, PHYH, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, POMT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PYGM, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH12, RDH5, REEP6, RGR, RGS9, RHO, RIMS2, RLBP1, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SCAPER, SDCCAG8, SGSH, SLC24A1, SLC38A8, SLC6A6, SNRNP200, SPATA7, SRD5A3, SSBP1, STN1, TIMM8A, TIMP3, TINF2, TMEM216, TMEM218, TMEM231, TMEM237, TOPORS, TPP1, TRAF3IP1, TREX1, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, WDPCP, WDR19, WHRN, ZFYVE26, ZNF408, ZNF423

Retinale Erkrankungen Tier 2: ADIPOR1, AHR, ASRGL1, MTRFR, CCT2, CEP19, CLCC1, CLUAP1, COQ5, CYP2R1, DHX38, DMD, ELOVL1, ESPN, EXOSC2, FRMD7, GDF6, IFT81, JAG1, KIF3B, LIG3, LRRC32, MAPKAPK3, MT-ATP6, MT-TH, MT-TL1, MT-TP, MT-TS2, OPN1SW, PDE6H, PGK1, POC5, POMGNT2, SLC66A1, RDH11, RTN4IP1, SAMD11, SAMD7, SEMA4A, SLC25A46, SLC37A3, SPG7, SPP2, SPTLC1, STX3, SUMF1, THRB, TTC21B, TTPA, UBAP1L, VWA8, ATXN7

Retinitis Pigmentosa Tier 1: ABCA4, ABHD12, AGBL5, AIPL1, AMACR, ARHGEF18, ARL2BP, ARL3, ARL6, BBS2, BEST1, C1QTNF5, PCARE, CFAP418, CDHR1, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CWC27, CYP4V2, DHDDS, EYS, FAM161A, FLVCR1, GUCA1B, GUCY2D, HGSNAT, HK1, IDH3A, IDH3B, IFT172, IMPDH1, IMPG1, IMPG2, KIZ, KLHL7, LRAT, MAK, MERTK, NEUROD1, NR2E3, NRL, OFD1, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, RAX2, RBP3, RBP4, RDH12, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, SAG, SCAPER, SGSH, SNRNP200, SPATA7, TOPORS, TRNT1, TTC8, TULP1, USH2A

Retinitis Pigmentosa Tier 2: AHR, CFAP20, DHX38, DYNC2H1, KIF3B, MVK, SEMA4A, SLC37A3, UBAP1L, VWA8, ADGRV1, BBS1, CFAP418, EXOSC2, ZNF408, PCARE, PRPH2, RCBTB1

Stäbchen-Zapfen Dystrophie: ABCA4, ADAM9, CFAP418, CDHR1, CERKL, CNGB3, CRX, EYS, FSCN2, GUCY2D, KCNV2, PDE6C, POC1B, PROM1, RAB28, RPE65, RPGRIP1, TULP1, DRAM2, SEMA4A, PCARE, GUCA1A, RIMS1, TTLL5, TLCD3B, PITPNM3, UNC119, RAX2, RPGR, CACNA1F, COG6, OPN1LW

Stargardt: ABCA4, ARMS2, BEST1, C1QTNF5, CDH3, CFH, CLN3, CNGB3, CRX, CTNNA1, DRAM2, ELOVL4, GUCA1A, IMPG1, IMPG2, IRX1, MFSD8, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTLL5

Syndromale Erkrankungen, Bindegewebsstörungen

Aicardi-Goutières-Syndrom: ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1

Autismus Tier: ABCD1, AGTR2, ANK3, ANKRD11, ARHGEF6, ARHGEF9, AVPR1A, BAZ2B, BCKDK, BCL11A, CACNA1D, CACNA1H, CACNA2D3, CHD1, CIC, CLIC2, CMIP, CNOT3, CNTN4, CNTN6, CNTNAP2, CRBN, CTNND2, CUX1, DDX3X, DEAF1, DHCR7, DIP2C, DLGAP2, DNMT3A, DPP6, EBF3, EED, EN2, EPB41L1, ERBIN, EXOC2, EXOC7, FBN1, FOXP1, GABRB3, GATM, GDI1, GIGYF2, GRIA1, GRIP1, HECW2, IGBP1, ILF2, INTS6, IRF2BPL, KAT2B, KDM5B, KDM6A, KDM6B, KMT2C, LAMC3, LAS1L, LEO1, MAGEL2, MAGT1,

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MBOAT7, MECP2, MED12, MED13L, MED23, MET, NCKAP1, NLGN3, NLGN4X, NRXN1, NSDHL, NTNG1, PHF3, PRPS1, PRSS12, PTCHD1, RAB39B, RANBP17, RELN, RIMS1, RPL10, SCN8A, SCN9A, SETD1B, SHANK2, SHROOM4, SLC6A1, SLC6A4, SLC9A9, SMARCA1, SMARCC2, SOBP, SPAST, SRCAP, SRSF11, ST3GAL3, SYP, TAOK2, TBL1XR1, TCF20, TECR, TMLHE, TNRC6B, TRIO, TSC1, TSC2, TSPAN7, UBN2, UPF3B, USP15, USP27X, USP7, WAC, WDFY3, ZC3H14, ZDHHC15, ZNF41, ZNF674, ZNF81

Autismus Tier 2: ACSL4, ADNP, ADSL, AFF2, AHDC1, ALDH5A1, ALG12, ALG3, ALG6, ANK2, ANKRD17, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARID1A, ARID1B, ARID2, ARX, ASH1L, ASXL1, ASXL2, ASXL3, ATP13A2, ATP6AP2, ATP7A, ATRX, AUTS2, BCAP31, BPTF, BRD4, BRSK2, BRWD3, CACNG2, CAMTA1, CASK, CC2D1A, CCDC22, CDH15, CDK13, CDK16, CHD2, CHD3, CHD7, CHD8, CLCN4, CNKSR2, CNOT1, CRADD, CREBBP, CSDE1, CSNK2A1, CTCF, CTNNB1, CUL3, CUL4B, DKC1, DLG3, DLG4, DPF2, DSCAM, DYRK1A, EBP, EFTUD2, EHMT1, EIF2S3, EIF3F, EZH2, FGD1, FGF12, FLNA, FMR1, FOLR1, FOXP2, FRMPD4, FTSJ1, GATAD2B, GK, GNAI1, GPC3, GRIA3, GRIK2, GRIN2B, H1-4, HCFC1, HDAC8, HEPACAM, HNRNPK, HNRNPR, HNRNPU, HOXA1, HPRT1, HUWE1, IDS, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KATNAL2, KCNC2, KCNH1, KDM5C, KIF1A, KMT2A, KMT2B, KMT2E, KMT5B, L1CAM, LINS1, MAN1B1, MAOA, MBD5, MBTPS2, MCPH1, MEF2C, MEIS2, MID1, MSL3, MYT1L, NAA10, NAA15, NBEA, NDP, NEXMIF, NHS, NONO, NR2F1, NR4A2, NSD1, NSD2, OCRL, OFD1, OPHN1, PACS1, PAK3, PGAP3, PHF21A, PHF6, PHF8, PHIP, PIGL, PIGN, PLP1, POGZ, PORCN, PPM1D, PPP2R1A, PPP2R5D, PQBP1, PTEN, PUF60, QRICH1, RAD21, RAI1, RERE, RPS6KA3, SATB2, SCN2A, SET, SETBP1, SETD5, SHANK3, SIN3A, SLC16A2, SLC2A1, SMAD4, SMARCA2, SMARCA4, SMC1A, SMC3, SMS, SNAP25, SON, SOX5, STAG1, SYN1, SYNGAP1, TANC2, TAOK1, TBR1, TCF7L2, TELO2, TRAPPC9, TRIM8, TRIP12, TRRAP, TUSC3, UBE2A, UBTF, USP9X, VPS13B, ZBTB18, ZC4H2, ZDHHC9, ZEB2, ZMIZ1, ZNF292, ZNF711

Bardet-Biedl-Syndrom: ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TTC8, WDPCP

CHARGE-Syndrom: CHD7

Coffin-Siris-Syndrom: ARID1A, ARID1B, BICRA, HDAC8, NIPBL, PHF6, PIGV, RAD21, SMARCA2, SMARCA4, SMC3, TBC1D24

Cornelia-de-Lange-Syndrom: HDAC8, NIPBL, RAD21, SMC3

Ehlers-Danlos Syndrom: ADAMTS2, ADAMTSL2, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, DSE, FKBP14, PLOD1, SLC39A13, AEBP1, COL5A1, COL5A2, TNXB, COL3A1

Epilepsie Tier 1: AFF3, AP2M1, ATP6V1A, CACNA1A, CACNA1E, CACNA1C, CACNA1D, CACNA1G, CACNA1I, CDK19, CELF2, CHD2, CHRNA4, CHRNB2, CNNM2, CSNK2B, CYFIP2, DEPDC5, DHDDS, DNM1, EEF1A2, FBXO28, FGF12, GABBR2, GABRA1, GABRA2, GABRA5, GABRB2, GABRB3, GABRG2, GNAO1, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2F, HCN1, HCN2, HECW2, HNRNPU, KCNB1, KCNC1, KCNC2, KCNQ2, KCNQ3, KCNT1, KCNT2, LGI1, MEF2C, NACC1, NPRL2, NPRL3, NTRK2, NUS1, PACS1, PACS2, PAK1, PHACTR1, PRRT2, RHOBTB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SETD1A, SETD1B, SIK1, SLC1A2, STX1B, STXBP1, TRAPPC4, TRIM8, VAMP2, WASF1, WDR37, YWHAG, ARX, CDKL5, CNKSR2, FGF13, OPHN1, PCDH19, SYN1, ABAT, ADARB1, ALDH7A1, AP3B2, ARV1, CACNA1B, CAD, CERS1, CNPY3, CNTNAP2, CSTB, CTNNA2, D2HGDH, DENND5A, DOCK7, EPM2A, FOLR1, FRRS1L, GOSR2, GOT2, GPAA1, ITPA, KCTD7, MDH2, MTHFS, NAPB, NHLRC1, NRROS, PARS2, PIGB, PLPBP, SCARB2, SLC13A5, SLC25A12, SLC38A3, SYNJ1, SZT2, TBC1D2B, TIAM1, TRAK1, UBA5, UGDH, UGP2, ZNF142

Epilepsie Tier 2: ACOX1, ACTL6B, AIMP2, AKT3, ALG8, ALPL, ANKRD11, ARF1, ARFGEF1, ARID1B, ASH1L, ASPA, ASXL3, ATN1, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5F1A, ATP6V0A1, ATP6V0C, BAP1, BOLA3, TET3, BRAF, BSCL2, CACNA1H, CCDC88C, CDC42BPB, CEP85L, CHD4, CHD5, CHRNA2, CIC, CLCN3, CLPB, CLTC, COG4, COG8, COL18A1, COL4A1, COL4A2, CPA6, CREBBP, CTU2, CUL3, CUX2, DEAF1, DHX16, DLL1, DNAJC5, DNM1L, DYNC1H1, DYRK1A, EFHC1, EFTUD2, EHMT1, EIF2AK2, EXT2, FAR1, FBXO11, FGFR3, FH, FOXG1, FRMD5, GABRB1, GABRD, GBA, GCH1, GFAP, GLI3, GLRA1, GLS, GLUD1, GNB1, GPHN, GRIA2, GRN, H3-3A, H3-3B, HEPACAM, HNRNPR, HRAS, HSPD1, IFIH1, IRF2BPL, KAT5, KAT8, KCNA1, KCNA2, KCNH1, KCNH5, KCNJ11, KCNK4, KCNMA1, KCNQ5, KIF1A, KIF2A, KIF5C, KMT2E, KRAS, LMBRD2, LMNB1, MACF1, MAF, MAP2K1, MAP2K2, MAST1, MBD5, MINPP1, MTHFR, MTOR, NARS1, NBEA, NCDN, NDE1, NEDD4L, NEUROD2, NR4A2, NSD1, NSF, OTX2, PAFAH1B1, PEX6, PIGT, PIK3R2, PNPT1, POLG2, PPP2CA, PPP3CA, PRODH, PRPF8, PSAP, PTEN, PUM1, PURA, RAB11B, RALA, RANBP2, RELN, RNF13, RNF2, RORA, RORB, RRM2B, RYR2, SAMHD1, SATB1, SATB2, SCO1, SCO2, SDHA, SERPINI1, SETBP1, SETD5, SGSH, SIX3, SLC12A5, SLC2A1, SLC6A1, SMARCA2, SMARCC2, SNAP25, SPTAN1, SPTBN1, STAG1, STARD7, SYNGAP1, TANC2, TBC1D24, TBL1XR1, TCF4, TMEM106B, TNPO2, TRAF7,

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TREX1, TRPM3, TRRAP, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, UBE3A, UFSP2, USP7, ZBTB18, ZEB2, ZMIZ1, ZMYM2, ALG13, ARHGEF9, ATP6AP2, ATP7A, ATRX, CASK, CLCN4, CUL4B, DCX, DDX3X, EIF2S3, FAM50A, FLNA, GLRA2, HCCS, HCFC1, HNRNP2, HPRT1, IKBKG, IQSEC2, MECP2, MED12, NDP, NDUFA1, NEXMIF, NSDHL, PDHA1, PIGA, RNF113A, SLC16A2, SLC35A2, SLC6A8, SLC9A6, SMC1A, SMS, TFE3, UBE2A, WDR45, XK, ZDHHC9, ADD1, AMT, ARF3, CAPRIN1, CSNK1G1, DROSHA, EMX2, GNAQ, HEATR5B, JAKMIP1, KCND2, KCTD3, KLHL20, PARP6, RAB11A, SCAF4, SCAMP5, SNX27, SPR, SYNCRIP, TNK2, TXNRD1, WNK3, AARS1, ABCA2, ADAM22, ADAR, ADAT3, ADGRG1, ADPRS, ADSL, AIMP1, ALDH5A1, ALG1, ALG11, ALG14, ALG3, ALG6, ALG9, ALKBH8, AMPD2, AP1G1, APC2, ARFGEF2, ARG1, ASAH1, ASNS, ATP6V0A2, BCKDHA, BCKDHB, BCS1L, BLOC1S1, BLTP1, BRAT1, BTBD, C12orf57, CACNA2D1, CACNA2D2, CARS2, CC2D2A, CCDC88A, CHKA, CLN3, CLN5, CLN6, CLN8, CNTN2, COG6, COG7, COLGALT1, COQ2, COQ4, COQ6, COQ9, COX10, COX15, CPLX1, CPSF3, CRPPA, CTSD, CTSF, CYP27A1, DBT, DDC, DEGS1, DHCR24, DHCR7, DHPS, DHX30, DIAPH1, DMXL2, DNAJC6, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPYD, DTYMK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF3F, EMC10, EML1, EPG5, ETHE1, EXOC7, EXOSC3, FARS2, FASTKD2, FBXL4, FCSK, FDFT1, FKRP, FKTN, FOXRED1, FUCA1, FUT8, GAD1, GALC, GALNT2, GAMT, GCSH, GFM1, GLB1, GLDC, GLUL, GLYCTK, GM2A, GNB5, GSS, GTPBP2, GTPBP3, GUF1, HACE1, HAX1, HERC2, HEXA, HEXB, HID1, HLCS, HMGCL, HOXA1, HPDL, HSD17B4, HTRA2, IER3IP1, KARS1, KATNB1, KCNJ10, KIFBP, KPTN, LARGE1, LARS1, LIAS, LIPT1, LIPT2, LMAN2L, LNP, LSS, LYST, MADD, MAGI2, MANBA, MBOAT7, MED17, MED27, MFF, MFSD8, MLC1, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MTR, NAGA, NARS2, NDUFA10, NDUFA2, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NGLY1, NRXN1, NSRP1, NUBPL, NUP214, OCLN, OGDHL, OTUD6B, OXR1, P4HTM, PAH, PCCA, PCCB, PCDH12, PCDHGC4, PCLO, PCYT2, PDHX, PDSS2, PET100, PEX1, PEX10, PEX12, PEX13, PEX19, PEX2, PEX3, PEX5, PEX7, PGM2L1, PHGDH, PIDD1, PIGC, PIGG, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGU, PIGW, PLA2G6, PLAA, PLCB1, PMM2, PMPCB, PNKP, POLG, POMGNT1, POMT1, POMT2, PPIL1, PPT1, PRICKLE1, PRMT7, PSAT1, PSPH, PTF1A, PTPN23, PTS, QARS1, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RALGAP1, RARS1, RARS2, RFT1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPIA, RTN4IP1, RTTN, RUSC2, SARS1, SEPSECS, SLC1A4, SLC25A1, SLC31A1, SLC35A1, SLC35A3, SLC39A8, SLC45A1, SLC5A6, SNIP1, SNORD118, SPATA5, SPATA5L1, SPTBN4, ST3GAL3, ST3GAL5, STAMBP, STRADA, SUCLA2, SUCLG1, SUOX, SURF1, TAF8, TANGO2, TBC1D20, TBCD, TDP2, TEO2, TIMM50, TMEM222, TMEM70, TMX2, TPP1, TRAPPC12, TRAPPC6B, TRIP13, TRPM6, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TUBGCP2, UBR7, UFC1, UFM1, UNC80, USP18, VARS1, VLDLR, VPS11, VPS50, WARS2, WDR45B, WDR62, WDR73, YIF1B, YIPF5, ZNF335

Joubert-Syndrom: AHI1, AIPL1, ARL13B, ARL6, ARMC9, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP290, CEP41, CPLANE1, CRB1, CRX, CSPP1, GUCY2D, HYL1, IFT140, IFT172, IFT27, IFT74, IMPDH1, INPP5E, INVS, IQCB1, KCNJ13, KIAA0586, KIAA0753, KIF7, LCA5, LRAT, LZTFL1, MERTK, MKS1, NMNAT1, NPHP1, NPHP3, NPHP4, OFD1, PIBF1, RD3, RDH12, RPE65, RPGRIP1, RPGRIP1L, SDCCAG8, SPATA7, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TTC21B, TTC8, TUBB4B, TULP1, TXNDC15, USP45

Kabuki-Syndrom: CHD7, EYA1, FLNB, IRF6, KDM6A, KMT2D, SIX1, SIX5

Loeys-Dietz-Syndrom: COL3A1, IPO8, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2

Marfan-Syndrom: FBN1, TGFB1, TGFB2, IPO8, COL1A2, FKBP14

Meckel-Gruber-Syndrom: B9D1, B9D2, CC2D2A, CEP290, CSPP1, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TCTN3, TMEM107, TMEM216, TMEM231, TMEM67, TXNDC15, WDPCP

Noonan Syndrom/RASopathie: BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2

Polyzystische Nierenerkrankung: ALG8, COL4A1, DNAJB11, DZIP1L, GANAB, GATM, HNF1B, LRP5, MUC1, NOTCH2, OFD1, PKD1, PKD2, PKHD1, PRKCSH, REN, SEC61A1, SEC63, TSC1, TSC2, TTC21B, UMOD, VHL

Primäre Ziliäre Dyskinesie: ACVR2B, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, CFAP300, CFAP45, CFAP52, CFAP53, CFC1, CFTR, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, MMP21, MNS1, NEK10, NME8, NODAL, ODAD1, ODAD2, ODAD3, ODAD4, PKD1L1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZIC3, ZMYND10

Rubenstein-Taybi-Syndrom: CREBBP, EP300, FGFR1, FGFR2, GLI3, HOXD13, SRCAP, TWIST1

Senior-Loken-Syndrom: CEP290, IQCB1, NPHP1, NPHP3, NPHP4, POC1B, SDCCAG8, TRAF3IP1, WDR19

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Smith-Lemli-Opitz-Syndrom: B9D2, BRAF, CBL, CC2D2A, CEP290, DHCR24, DHCR7, EBP, FDF1, GLI3, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MKS1, MRAS, NF1, NPHP3, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RPGRIP1L, RRAS2, SC5D, SHOC2, SOS1, SOS2, SPRED1, SPRED2, TCTN2, TMEM216, TMEM231, TMEM67, TXNDC15

Tumordispositionen

Tumordisposition breit: AKT1, APC, ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CDKN1B, CDKN2A, CHEK2, CTNNA1, DDB2, DICER1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FH, MAP2K1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, POLD1, POLE, POLH, PTCH1, PMS2, PTEN, PTPN11, RAD51C, RAD51D, SLX4, SMARCA4, SMARCB1, STK11, SUFU, TOP3A, TP53, TRIM37, UBE2T, WRN, WT1, XPA, XPC, XRCC2, ACD, ALK, BAP1, BMPR1A, BUB1B, CDC73, CDK4, CDKN1C, CTC1, DKC1, DOCK8, ELANE, ELP1, EXT1, EXT2, FLCN, GATA1, GBA, GNAS, GPC3, GPR161, HRAS, KRAS, LIG4, MAD2L2, MAX, MEN1, NAF1, NF2, NHP2, NOP10, NRAS, NSD1, PARN, POT1, PRKAR1A, RAF1, RB1, RECQL4, REST, RET, RIT1, RMRP, RNF43, RPA1, RPL11, RPL15, RPL23, RPL26, RPL31, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SMAD4, SOS1, STN1, TERC, TERT, TINF2, TMEM127, TRIP13, TSC1, TSC2, VHL, WRAP53, AXIN2, GREM1, MSH3, BRAF, HOXB13, LZTR1, PDGFRB, SPRED1, MET, PDGFRA, EZH2, COL3A1, CYLD, FBN1, IDH2, DLST, MDH2, SLC25A11, GPR101, IDH1, AIP, CASR, DNMT3A, EGLN1, EPAS1, H3-3B, KIF1B, MERTK, SUCLG2, ANKRD26, CBL, CEBPA, CTR9, DDX41, DIS3L2, DNAJC21, ETV6, FAS, GATA2, HAX1, IKZF1, ITK, KIT, MAP2K2, NYNRIN, PAX5, PHOX2B, PPP1CB, PRF1, RAD51, RUNX1, SBDS, SERPINA1, SH2D1A, STAT3, TRIM28, WAS

Birt-Hogg-Dubé Syndrom: BAP1, CDC73, CDKN1B, CHEK2, COL3A1, CYLD, EPCAM, FBN1, FH, FLCN, IDH2, MAX, MEN1, MET, MLH1, MSH2, MSH6, PMS2, PRKAR1A, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, TSC1, TSC2, VHL

Blasen- und Harnleiterkrebs Tier 1: EPCAM, MLH1, MSH2, MSH6, PMS2, BAP1, MUTYH, NTHL1

Blasen- und Harnleiterkrebs Tier 2: NF1, SMARCA4, SMARCB1, STK11, TP53, FH

Cowden Syndrom: AKT1, APC, ATM, AXIN2, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDKN1B, CDKN2A, CHEK2, DDB2, DICER1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FH, FLCN, GREM1, MET, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, POLH, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SMAD4, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, XPA, XPC

Endometriumkarzinom: FH, MLH1, MSH2, MSH6, PMS2, PTEN, EPCAM, MUTYH, NTHL1, POLD1, POLE, SMAD4

Familiärer Brust- und Eierstockkrebs (Therapierelevanz): BRCA1, BRCA2

Familiärer Brust- und Eierstockkrebs Tier 1: ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53

Familiärer Brust- und Eierstockkrebs Tier 2: AKT1, APC, BLM, CDKN1B, CDKN2A, CTNNA1, DDB2, DICER1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FH, MAP2K1, MUTYH, NTHL1, POLD1, POLE, POLH, PTCH1, PTPN11, SLX4, SMARCA4, SMARCB1, SUFU, TOP3A, TRIM37, UBE2T, WRN, WT1, XPA, XPC, XRCC2

GIST: KIT, PDGFRA, SDHA, SDHB, SDHC, SDHD

Gorlin-Goltz Syndrom: AKT1, APC, BAP1, BUB1B, CDKN1C, DDB2, DICER1, ELP1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EZH2, GPC3, GPR161, HRAS, NBN, NF1, NSD1, PALB2, PDGFRB, POLH, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, RMRP, SMARCA4, SMARCB1, SOS1, SUFU, TRIP13, TSC1, TSC2, XPA, XPC

Hereditäres Phäochromozytom-Paragangliom-Syndrom: DLST, EGLN1, EPAS1, FH, H3-3B, KIF1B, MAX, MDH2, MEN1, MERTK, MET, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SUCLG2, TMEM127, TP53, VHL

Solide kindliche Tumore Tier 1: PTPN11, RAF1, SOS1, BRAF, PDGFRB, EPCAM, MLH1, MSH2, MSH6, PMS2, NF1, NF2, SMARCB1, APC, BMPR1A, PTEN, STK11, ALK, ATM, BRCA1, BRCA2, BRIP1, CDKN1B, CDKN1C, DICER1, ELP1, GPC3, GPR161, HRAS, KRAS, MEN1, NRAS, PALB2, POLH, PTCH1, RB1, RECQL4, REST, RET, SHOC2, SMARCA4, SUFU, TRIM37, TRIP13, TSC1, TSC2, VHL, WRN, WT1, XPA, XPC, TP53, CBL,

CTR9, DIS3L2, RUNX1, SERPINA1, TRIM28, BLM, BUB1B, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, SLX4, UBE2T, TOP3A
Solide kindliche Tumore Tier 2: GNAS, RIT1, MAP2K1, PRKAR1A, LZTR1, SPRED1, CHEK2, MUTYH, NTHL1, POLD1, POLE, SMAD4, ACD, AKT1, BAP1, BARD1, CDC73, CDH1, CDK4, CDKN2A, CTC1, DKC1, ELANE, EXT1, EXT2, FH, FLCN, GATA1, GBA, LIG4, MAD2L2, MAX, NAF1, NBN, NHP2, NOP10, NSD1, PARN, POT1, RAD51C, RAD51D, RMRP, RPL11, RPL15, RPL23, RPL26, RPL31, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, SEC23B, STN1, TERC, TERT, TINF2, WRAP53, XRCC2, ANKRD26, CEBPA, DDX41, DNAJC21, ETV6, EZH2, FAS, GATA2, HAX1, IKZF1, ITK, KIT, MAP2K2, MET, NYNRIN, PAX5, PHOX2B, PPP1CB, PRF1, RAD51, SBDS, SH2D1A, STAT3, WAS
Li-Fraumeni-Syndrom: CHEK2, ACD, AKT1, ALK, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CTC1, DDB2, DICER1, DKC1, DOCK8, ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FH, FLCN, GATA1, GBA, GNAS, GPC3, GPR161, HRAS, KRAS, LIG4, MAD2L2, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NAF1, NBN, NF1, NF2, NHP2, NOP10, NRAS, NSD1, NTHL1, PALB2, PARN, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD51C, RAD51D, RAF1, RB1, RECQL4, REST, RET, RIT1, RMRP, RNF43, RPA1, RPL11, RPL15, RPL23, RPL26, RPL31, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, STK11, STN1, SUFU, TERC, TERT, TINF2, TMEM127, TP53, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2
Lynch-Syndrom: MLH1, PMS2, MSH6, MSH2, EPCAM
Magenkrebs: AAGAB, APC, ATM, BLM, BMPR1A, BRCA2, BUB1, BUB1B, BUB3, CASP10, CDH1, CDKN2A, CEP57, CHEK2, COL14A1, ERBB2, FGFR2, IL1B, IL1RN, IRF1, KLF6, KRAS, MDM2, MSH3, MUTYH, PIK3CA, POLD1, POLE, RPS20, SEMA4A, STK11, TP53, TRIP13, CTNNA1, EPCAM, MLH1, MSH2, MSH6, PDGFRA, PMS2, PTPN11, SMAD4
Medulloblastom: APC, BRCA2, DICER1, ELP1, GPR161, PALB2, PTCH1, RET, SEC23B, SUFU, TP53, GPC3, NBN
Melanom: BAP1, CDK4, CDKN2A, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, GBA, GNAS, MITF, POLH, POT1, PTEN, RB1, RECQL4, TERF2IP, WRN, XPA, XPC, ACD, BRCA2, TERT
Multiple endokrine Neoplasie Tier 1: CDKN1B, MEN1, RET
Multiple endokrine Neoplasie Tier 2: CDC73, FH, MAX, NF1, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, UBE2T, TP53, AIP, CASR, DLST, DNMT3A, MDH2, SLC25A11, VHL
Multiple Exostosen: APC, EXT1, EXT2, GATA1, GNAS, GPR101, IDH1, PRKAR1A, PTPN11, RB1, RECQL4, RPL11, RPL15, RPL23, RPL26, RPL31, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, TP53, WRN
Neurofibromatose / Schwannomatose Tier 1: NF1, NF2, PRKAR1A, SMARCB1, LZTR1, SPRED1
Neurofibromatose / Schwannomatose Tier 2: GNAS, PTPN11, RAF1, RIT1, SOS1, EPCAM, MLH1, MSH2, MSH6, PMS2, BRAF, HOXB13, MAP2K1, PDGFRB, BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, KITLG, PALB2, SLX4
Nierenkarzinom: APC, BAP1, CDC73, CDKN1B, CHEK2, DDB2, DICER1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FH, FLCN, MET, MLH1, MSH2, MSH6, NRAS, PMS2, POLH, PRKAR1A, PTEN, RAF1, SDHA, SDHAF2, SDHB, SDHC, SDHD, STK11, TMEM127, TP53, TSC1, TSC2, VHL, XPA, XPC
Pankreaskarzinom Tier 1: ATM, BRCA1, BRCA2, CDKN2A, MLH1, MSH2, MSH6, PMS2, PALB2, STK11, TP53
Pankreaskarzinom Tier 2: APC, BMPR1A, EPCAM, PRSS1, PTEN, SMAD4, SPINK1, VHL
Peutz-Jeghers Syndrom: APC, ATM, AXIN2, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, PRKAR1A, PTEN, RAD51C, RAD51D, RNF43, SEC23B, SMAD4, STK11, TP53
Polyposis Tier 1: APC, BMPR1A, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, AXIN2, GREM1, EPCAM, MLH1, MSH2, MSH6, PMS2
Polyposis Tier 2: EPCAM, MLH1, MSH2, MSH6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, SEC23B, SLX4, UBE2T, XRCC2, MSH3
Sarkoma Tier 1: MTAP, PDGFRA, RECQL4, SQSTM1, TBXT, TP53, EXT1, EXT2, NF1

Sarkoma Tier 2: APC, ATM, ATR, BLM, BRCA2, BUB1B, CDKN1C, ERCC2, FANCC, FH, HRAS, KIT, MLH1, MSH2, MSH6, NBN, PMS2, RB1, SDHA, SDHB, SDHD, SMARCA4, WRN
Schilddrüsenkarzinom Tier 1: APC, CHEK2, CDC73, CDKN1B, DICER1, FLCN, POLH, PRKAR1A, PTEN, RET, TSC1, TSC2, WRN, TP53
Schilddrüsenkarzinom Tier 2: XPA, XPC, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, MUTYH, GNAS, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5
Tuberöse Sklerose Tier 1: TSC1, TSC2
Tuberöse Sklerose Tier 2: CHEK2, EPCAM, MLH1, MSH2, MSH6, PMS2, BAP1, CDC73, CDKN1B, DDB2, DICER1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FH, FLCN, MAX, MEN1, NF1, NF2, NSD1, POLH, POT1, PRKAR1A, PTCH1, PTEN, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SUFU, TMEM127, VHL, XPA, XPC, TP53, AIP, DNMT3A, KIT, MET
Von-Hippel-Lindau Syndrom: POLD1, POLE, DDB2, ERCC3, ERCC4, ERCC5, XPA, XPC, POLH, ERCC1, PTEN, CHEK2, DICER1, MLH1, MSH2, MSH6, PMS2, EPCAM, STK11, TP53, CDKN1B, FH, NF1, MAX, MEN1, RET, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TSC1, TSC2, BAP1, CDC73, DLST, FLCN, MDH2, MET, PRKAR1A, SDHA, SLC25A11, VHL

Weitere Erkrankungen

Erbliche Blutungserkrankungen Tier 1: ABCG5, ABCG8, ACTB, ACTN1, ACVRL1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S6, CDC42, CHST14, COL1A1, COL5A1, COL5A2, CYCS, DIAPH1, DTNBP1, ENG, ETV6, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FERMT3, FGA, FGB, FGG, FLI1, FLNA, FYB1, GATA1, GBA, GFI1B, GGCX, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRG, IKZF5, ITGA2B, ITGB3, KDSR, KLKB1, KNG1, LMAN1, LYST, MCFD2, MECOM, MPIG6B, MPL, MYH9, NBEA, NBEAL2, P2RY12, PLA2G4A, PLAT, PLAU, PLG, PROC, PROS1, PTPN11, RASGRP2, RBM8A, RNU4ATAC, RUNX1, SERPINC1, SERPIND1, SERPINE1, SERPINF2, SLC45A2, SLFN14, SMAD4, SRC, STIM1, STXBP2, TBXA2R, TBXAS1, THBD, THPO, TUBB1, VIPAS39, VKORC1, VPS33B, VWF, WAS
Erbliche Blutungserkrankungen Tier 2: LAT, ORAI1, PTPRJ, SLC35A1, TPM4
Leukodystrophie Tier 1: AARS1, AARS2, ABCD1, ALDH3A2, APP, ARSA, AUH, CLCN2, COL4A1, COL4A2, CSF1R, CTSA, CYP27A1, DARS1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, GALC, GBE1, GFAP, GJA1, GJB1, GJC2, GLA, GLB1, GSN, HEPACAM, HEXA, HMGCL, HTRA1, ITM2B, L2HGDH, LAMB1, LARS2, LIG3, LMNB1, MARS1, MCOLN1, MTHFR, NOTCH3, OCRL, PAH, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PLP1, POLR1C, POLR3A, POLR3B, PRNP, PSAP, PSEN1, PTEN, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RPS6KA3, SAMHD1, SNORD118, SPG11, SPG21, TREM2, TREX1, TTR, TUBB4A, TYMP, TYROBP, ZFYVE26, MLC1
Leukodystrophie Tier 2: CST3, CTC1, EARS2, GCDH, KIF5A, MAL, NPC1, PSEN2, TPP2
Okulärer und okulokutaner Albinismus: GPR143, HPS1, HPS3, HPS4, HPS5, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1
Osteogenesis imperfecta Tier 1: ATP6V0A2, B4GALT7, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL5A1, PLOD1, TNXB, ALPL, B3GALT6, BMP1, CASR, COL12A1, CREB3L1, CRTAP, DSPP, TENT5A, FKBP10, GORAB, IFITM5, KDELR2, LRP5, MESD, NBAS, NOTCH2, P3H1, P4HB, PLOD2, PLS3, PPIB, PYCR1, SEC24D, SERPINF1, SERPINH1, SLC26A2, SP7, SPARC, TAPT1, TMEM38B, TRPV6, UNC45A, WNT1
Osteogenesis imperfecta Tier 2: COPB2, MBTPS2, NUDT6, SGMS2, SUCO
Primäre Ovarialinsuffizienz Tier 1: MSH4, SYCE1, C14orf39, AARS2, BMP15, CLPP, CYP17A1, CYP19A1, EIF2B5, FANCM, FOXL2, FSHB, FSHR, GALT, GGPS1, HFM1, HSD17B4, LARS2, MCM8, MCM9, NOBOX, NR5A1, PMM2, POLG, SOHLH1, STAG3, TWNK, HARS2
Primäre Ovarialinsuffizienz Tier 2: EIF2B4, GDF9, NOG, NUP107, POU5F1, RCBTB1, SOHLH2, BMPR1B, BUB1B, DACH2, EIF4ENIF1, FIGLA, LMNA, MSH5, NANOS3, PGRMC1, POF1B, POLR2C, POLR3H, PSMC3IP, SGO2, SYCP2L, KHDRBS1
Primäre Ovarialinsuffizienz Tier 3: BLM, ATM, EIF2B2, FANCA, FANCG, HSF2BP, NBN, RECQL4, WRN, ACAD9, ACSL6, ADAMTS19, AGTR2, AIFM1, AIRE, AKT1, ALOX12B, ANKRD22, ANKRD31, AR, BAX, BCKDHB, BCORL1, BHLHB9, BNC1, BRCA2, BRSK1, CARD11, CCBE1, CCNH, CHM, CITED2, COX10, CPEB1, VCAN, CTNNA3, CYP2E1, DAZL, DHFR, DLX5, DLX6, DMC1, DNAH5, SEM1, DUSP22, ERCC6, ESR1, EXO1,

Erstellung: 06.11.2024 Lißewski, Christina
 Prüfung: 08.11.2024 Dombrowsky, Gregor
 Genehmigung: 12.11.2024 Hitz, Marc-Phillip

Version: 1.0
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FANCL, AFF2, FOXO1, FOXO3, GJA1, GNAS, HAO2, HK3, HSD3B2, INHA, LAMC1, LHB, LHCGR, MEF2C, MRPS22, NAIP, NR2F1, NRXN1, NUPR1, NXF5, PRKN, PCDH19, PCMT1, PMAIP1, PMM1, POR, PRDM9, PTEN, BBS9, RAD51, SF1, SHOX, SPATA22, SPIDR, SSBP2, STAMBPL1, STAR, STS, TGFBR3, TP63, TSPAN7, UTP14A, VCX, WDR62, WNT4, WT1, XIST, XPNPEP2, XRCC2, YBX2, ZFX