

Liste der im flexiblen Bereich akkreditierten Verfahren

Stand: 07.04.2026

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Medizinisches Versorgungszentrum am Klinikum Oldenburg GmbH

Medizinische Genetik

Rahel-Straus-Str. 10, 26133 Oldenburg

Untersuchungsart:

Molekularbiologische Untersuchungen [Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	Letzte Aktualisierung
Angeborene Herzfehler breit (virtuelles Panel, Illumina PCR-Free Library Prep) (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, TGFBR1, TGFBR2, ZEB2, ARID1A, ASXL1, BCOR, BRAF, CDK13, CFC1, CHD4, CREBBP, EHMT1, EP300, EVC, EVC2, FLT4, HRAS, KDR, LZTR1, MRAS, NF1, PPP1CB, PRKD1, RAI1, RBF2, RASGEF1B, RASGEF2, RASGEF3, RASGEF4, RASGEF5, RASGEF6, RASGEF7, RASGEF8, RASGEF9, RASGEF10, RASGEF11, RASGEF12, RASGEF13, RASGEF14, RASGEF15, RASGEF16, RASGEF17, RASGEF18, RASGEF19, RASGEF20, RASGEF21, RASGEF22, RASGEF23, RASGEF24, RASGEF25, RASGEF26, RASGEF27, RASGEF28, RASGEF29, RASGEF30, RASGEF31, RASGEF32, RASGEF33, RASGEF34, RASGEF35, RASGEF36, RASGEF37, RASGEF38, RASGEF39, RASGEF40, RASGEF41, RASGEF42, RASGEF43, RASGEF44, RASGEF45, RASGEF46, RASGEF47, RASGEF48, RASGEF49, RASGEF50, RASGEF51, RASGEF52, RASGEF53, RASGEF54, RASGEF55, RASGEF56, RASGEF57, RASGEF58, RASGEF59, RASGEF60, RASGEF61, RASGEF62, RASGEF63, RASGEF64, RASGEF65, RASGEF66, RASGEF67, RASGEF68, RASGEF69, RASGEF70, RASGEF71, RASGEF72, RASGEF73, RASGEF74, RASGEF75, RASGEF76, RASGEF77, RASGEF78, RASGEF79, RASGEF80, RASGEF81, RASGEF82, RASGEF83, RASGEF84, RASGEF85, RASGEF86, RASGEF87, RASGEF88, RASGEF89, RASGEF90, RASGEF91, RASGEF92, RASGEF93, RASGEF94, RASGEF95, RASGEF96, RASGEF97, RASGEF98, RASGEF99, RASGEF100)	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_CHD/AHF_breit_Tier 1 und Kardio_CHD/AHF_breit_Tier 2, Pipeline V.2	07.03.2025

HDAC8, HEY2, HNRNPK, HSPA9, HYAL2, HYLS1, IDUA, IGFBP7, IPO8, IRX4, KANSL1, KAT6A, KAT6B, KDM5B, KMT2A, KYNU, LAMA4, LEFTY1, LEFTY2, LTBP2, MAP3K7, MAPK1, MAPKAPK5, 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); SNV, CNV				
Angeborene Herzfehler nicht syndromal (virtuelles Panel, Illumina PCR-Free Library Prep) (ACTC1, ELN, FBN1, FLNA, GATA4, GATA6, GDF1, GJA1, JAG1, MED13L, NKX2-5, NODAL, NOTCH1, NOTCH2, NR2F2, SOS1, TAB2, TBX1, TBX20, TBX5, TGFBR1, TGFBR2, CFC1, FLT4, KDR, RBFOX2, SALL1, SLC2A10, ACVR2B, CITED2, CRELD1, GATA5, MYH6, NKX2-6, SMAD6, TLL1, ZFPM2, 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); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_CHD/AHF_nicht-syndromal_Tier 1 und Kardio_CHD/AHF_nicht-syndromal_Tier 2, Pipeline V.2	07.03.2025
Arrhythmien (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ACTC1, ACTN2, ALPK3, BAG3, CACNA1C, CALM1, CALM2, CALM3, CASQ2, CDH2, CSRP3, DES, DMD, DOLK, DSC2, DSP, EMD,	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_Arrhythmia_v2025_Tier1 und Kardio_Arrhythmia_v2025_Tier2, Pipeline V.2	02.04.2026

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, 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, TECRL, TRDN, TRPM4, TULP3); SNV				
Arrhythmien (virtuelles Panel, Illumina PCR-Free Library Prep) (ACTC1, ACTN2, ALPK3, BAG3, CACNA1C, CALM1, CALM2, CALM3, CASQ2, CDH2, CSR3, 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, 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, TECRL, TRDN, TRPM4, TULP3); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_Arrhythmia_v2025_Tier1 und Kardio_Arrhythmia_v2025_Tier2, Pipeline V.2	02.04.2026
ARVC (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ANK2, CAV3, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MYH7,	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_ARVC_v2025_Tier1 und Kardio_ARVC_v2025_Tier2, Pipeline V.2	13.08.2025

MYL3, PKP2, PLN, SCN5A, TGFB3, TJP1, TMEM43, TTN); SNV				
ARVC (virtuelles Panel, Illumina PCR-Free Library Prep) (ANK2, CAV3, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MYH7, MYL3, PKP2, PLN, SCN5A, TGFB3, TJP1, TMEM43, TTN); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_ARVC_v2025_Tier1 und Kardio_ARVC_v2025_Tier2, Pipeline V.2	13.08.2025
Autoinflammation (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ADA2, ARD14, IL17RA, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLR4, NLRP3, NOD2, OGFRL1, OTULIN, PLCG2, PRF1, PSMB4, PSMB8, PSMB9, PSTPIP1, RBCK1, SH3BP2, SLC29A3, TMEM173, TNAIP3, TNFRSF1A, UBA1); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Autoinflammation_v2025_Tier1 und Kardio_Autoinflammation_v2025_Tier2, Pipeline V.2	27.03.2026
Autoinflammation (virtuelles Panel, Illumina PCR-Free Library Prep) (ADA2, ARD14, IL17RA, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLR4, NLRP3, NOD2, OGFRL1, OTULIN, PLCG2, PRF1, PSMB4, PSMB8, PSMB9, PSTPIP1, RBCK1, SH3BP2, SLC29A3, TMEM173, TNAIP3, TNFRSF1A, UBA1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Autoinflammation_v2025_Tier1 und Kardio_Autoinflammation_v2025_Tier2, Pipeline V.2	27.03.2026
Bardet-Biedl-Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TTC8, WDPCP); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Syndrom_Bardet-Biedl_v2026, Pipeline V.2	09.03.2026
Bardet-Biedl-Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, CFAP418, IFT172, IFT27, IFT57, IFT74, INPP5E, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC21B, TTC8, WDPCP); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Bardet-Biedl_v2026, Pipeline V.2	09.03.2026
Blasen- und Harnleiterkrebs (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (BAP1, EPCAM, FH, MLH1, MSH2, MSH6, MUTYH, NF1,	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Blasen+Harnleiterkrebs_v.2025_Tier1, und Tumordisp_Blasen+Harnleiterkrebs_v.2025_Tier2 Pipeline V.2	28.07.2025

NTHL1, PMS2, SMARCA4, SMARCB1, STK11, TP53); SNV				
Blasen- und Harnleiterkrebs (virtuelles Panel, Illumina PCR-Free Library Prep) (BAP1, EPCAM, FH, MLH1, MSH2, MSH6, MUTYH, NF1, NTHL1, PMS2, SMARCA4, SMARCB1, STK11, TP53); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Blasen+Harnleiterkrebs_v.2025_Tier1, und Tumordisp_Blasen+Harnleiterkrebs_v.2025_Tier2 Pipeline V.2	28.07.2025
Brugada-Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (CACNA1C, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNH2, SCN1B, SCN3B, SCN5A, TRPM4); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_Brugada_v2025_Tier1 und Kardio_Brugada_v2025_Tier2, Pipeline V.2	15.08.2025
Brugada-Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (CACNA1C, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNH2, SCN1B, SCN3B, SCN5A, TRPM4); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_Brugada_v2025_Tier1 und Kardio_Brugada_v2025_Tier2, Pipeline V.2	15.08.2025
Cowden Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (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, PIK3CA, 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); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Cowden_v2025, Pipeline V.2	14.07.2025
Cowden Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (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, PIK3CA, PMS2, POLD1, POLE, POLH, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Cowden_v2025, Pipeline V.2	14.07.2025

SEC23B, SMAD4, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, XPA, XPC); SNV, CNV				
CPVT (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (CALM1, CALM2, CALM3, CASQ2, RYR2, TECRL, TRDN); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_CPVT_v2025_Tier1only, Pipeline V.2	15.08.2025
CPVT (virtuelles Panel, Illumina PCR-Free Library Prep) (CALM1, CALM2, CALM3, CASQ2, RYR2, TECRL, TRDN); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_CPVT_v2025_Tier1only, Pipeline V.2	15.08.2025
Darmkrebs (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (AKT1, APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, FLCN, GALNT12, GREM1, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SEC23B, SMAD4, STK11, TP53); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Darmkrebs_v.2025_Tier 1 und Tumordisp_Darmkrebs_v.2025_Tier 2, Pipeline V.2	28.07.2025
Darmkrebs (virtuelles Panel, Illumina PCR-Free Library Prep) (AKT1, APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, FLCN, GALNT12, GREM1, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SEC23B, SMAD4, STK11, TP53); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Darmkrebs_v.2025_Tier 1 und Tumordisp_Darmkrebs_v.2025_Tier 2, Pipeline V.2	28.07.2025
DCM (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ABCC9, ACTC1, ACTN2, ANK2, ANKRD1, BAG3, CDH2, CRYAB, CSRP3, CTF1, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EPG5, EYA4, FKRP, FKTN, FLII, FLNC, GATA6, GATAD1, IDH2, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYLK3, MYPN, MYZAP, NEBL, NEXN, NKX2-5, NRAP, PKP2, PLN, PRDM16, PSEN1, RAF1, RBM20, RHBDF1, RPL3L, RRAGC, RRAGD, RYR2, SCN1B, SCN5A, SGCD, SLC6A6, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_DCM_v2025_Tier1 und Kardio_DCM_v2025_Tier2, Pipeline V.2	15.08.2025
DCM (virtuelles Panel, Illumina PCR-Free Library Prep) (ABCC9, ACTC1, ACTN2, ANK2, ANKRD1, BAG3, CDH2, CRYAB, CSRP3, CTF1, DES, DMD, DOLK, DSC2,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_DCM_v2025_Tier1 und Kardio_DCM_v2025_Tier2, Pipeline V.2	15.08.2025

DSG2, DSP, DTNA, EPG5, EYA4, FKR, FKTN, FLII, FLNC, GATA6, GATAD1, IDH2, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYLK3, MYPN, MYZAP, NEBL, NEXN, NKX2-5, NRAP, PKP2, PLN, PRDM16, PSEN1, RAF1, RBM20, RHBDF1, RPL3L, RRAGC, RRAGD, RYR2, SCN1B, SCN5A, SGCD, SLC6A6, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL); SNV, CNV				
Ehlers-Danlos Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ADAMTS2, ADAMTSL2, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, DSE, FKBP14, PLOD1, SLC39A13, AEBP1, COL5A1, COL5A2, TNXB, COL3A1); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Syndrom_Ehlers-Danlos_v.2025 T1+2 Pipeline V.2	02.04.2025
Ehlers-Danlos Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (ADAMTS2, ADAMTSL2, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, DSE, FKBP14, PLOD1, SLC39A13, AEBP1, COL5A1, COL5A2, TNXB, COL3A1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Ehlers-Danlos_v.2025 T1+2 Pipeline V.2	02.04.2025
Endometriumkarzinom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (EPCAM, FH, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_EndometriumCA_v2025_only1Tier, Pipeline V.2	18.07.2025
Endometriumkarzinom (virtuelles Panel, Illumina PCR-Free Library Prep) (EPCAM, FH, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_EndometriumCA_v2025_only1Tier, Pipeline V.2	18.07.2025
Epilepsie (virtuelles Panel, Illumina PCR-Free Library Prep) (AARS, AASS, ABAT, ABCA2, ACOX1, ACTL6B, ADAM22, ADAR, ADARB1, ADAT3, ADD1, ADGRG1, ADPRHL2, ADSL, AFF3, AGO1, AIMP1, AIMP2, AKT3, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG13, ALG14, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANKRD11, AP1G1, AP2M1, AP3B2, APC2, ARF1,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Epilepsy_v2026_Tier1 und Syndrom_Epilepsy_v2026_Tier2, Pipeline V.2	30.09.2025

<p>ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGEF9, ARID1B, ARV1, ARX, ASAH1, ASH1L, ASNS, ASPA, ASXL3, ATN1, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5A1, ATP5O, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATRX, BAP1, BCKDHA, BCKDHB, BCS1L, BLOC1S1, BOLA3, BRAF, BRAT1, BSCL2, BTB, C12orf57, C2orf69, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D1, CACNA2D2, CAD, CAMSAP1, CAPRIN1, CARS2, CASK, CC2D2A, CCDC88A, CCDC88C, CCM2, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CHD2, CHD4, CHD5, CHKA, CHRNA2, CHRNA4, CHRN2, CIC, CLCN3, CLCN4, CLN3, CLN5, CLN6, CLN8, CLPB, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP2, COG4, COG6, COG7, COG8, COL18A1, COL4A1, COL4A2, COL4A3BP, COLGALT1, COQ2, COQ4, COQ6, COQ9, COX10, COX15, CPA6, CPLX1, CPSF3, CREBBP, CRELD1, CSNK1G1, CSNK2B, CSTB, CTNNA2, CTSD, CTSF, CTU2, CUL3, CUL4B, CUX1, CUX2, CYFIP2, CYP27A1, D2HGDH, DBT, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DEPDC5, DHCR24, DHCR7, DHDDS, DHPS, DHX16, DHX30, DIAPH1, DLL1, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPYD, DROSHA, DTYMK, DYNC1H1, DYRK1A, EARS2, EEF1A2, EFHC1, EFTUD2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EMC10, EML1, EMX2, EPG5, EPM2A, ESAM, ETHE1, EXOC7, EXOSC3, EXT2, FAM50A, FAR1, FARS2, FASTKD2, FBXL4, FBXO11, FBXO28, FDFT1, FGF12, FGF13, FGFR3, FH, FKRP, FKTN, FLNA, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FUCA1, FUK, FUT8, FZR1, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNT2, GAMT, GBA, GCH1, GCSH, GFAP, GFM1, GLB1, GLDC, GLI3, GLRA1, GLRA2, GLS, GLUD1, GLUL, GLYCTK, GM2A, GNAO1, GNAQ, GNB1, GNB5, GOSR2, GOT2, GPAA1, GPHN,</p>				
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<p>GRIA2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GSS, GTPBP2, GTPBP3, GUF1, H3F3A, H3F3B, HACE1, HAX1, HCCS, HCFC1, HCN1, HCN2, HDAC4, HEATR5B, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HID1, HLCS, HMGCL, HNRNPH2, HNRNPR, HNRNPU, HOXA1, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA2, IER3IP1, IFIH1, IKBKG, IQSEC2, IRF2BPL, ISPD, ITPA, JAKMIP1, KARS, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM6B, KIAA1109, KIF1A, KIF1BP, KIF2A, KIF5C, KLHL20, KMT2E, KPTN, KRAS, KRIT1, LARGE1, LARS, LETM1, LGI1, LIAS, LIPT1, LIPT2, LMAN2L, LMBRD2, LMNB1, LNPB, LSS, LYST, MACF1, MADD, MAF, MAGI2, MANBA, MAP2K1, MAP2K2, MAST1, MAST3, MAST4, MBD5, MBOAT7, MDH2, MECP2, MED11, MED12, MED17, MED27, MEF2C, MFF, MFSB8, MINPP1, MLC1, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MTHFR, MTHFS, MTOR, MTR, NACC1, NAGA, NAPB, NARS, NARS2, NBEA, NCDN, NDE1, NDP, NDUFA1, NDUFA10, NDUFA2, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEUROD2, NEXMIF, NGLY1, NHLRC1, NPRL2, NPRL3, NR4A2, NRROS, NRXN1, NSD1, NSDHL, NSF, NSRP1, NTRK2, NUBPL, NUP214, NUS1, OCLN, OGDHL, OPHN1, OTUD6B, OTUD7A, OTX2, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PARP6, PARS2, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCLO, PCYT2, PDCD10, PDHA1, PDHX, PDSS2, PET100, PEX1, PEX10, PEX12, PEX13, PEX19, PEX2, PEX3, PEX5, PEX6, PEX7, PGM2L1, PHACTR1, PHGDH, PI4K2A, PID1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGW, PIK3R2, PLA2G6, PLAA, PLCB1, PLPBP, PMM2, PMPCB, PNKP, PNPO,</p>				
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<p>PNPT1, POLG, POLG2, POMGNT1, POMT1, POMT2, PPFIBP1, PPIL1, PPP2CA, PPP3CA, PPT1, PRICKLE1, PRMT7, PRODH, PRPF8, PRRT2, PSAP, PSAT1, PSPH, PTC3, PTEN, PTF1A, PTPN23, PTS, PUM1, PURA, QARS, QDPR, RAB11A, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAC3, RALA, RALGAPA1, RANBP2, RARS, RARS2, RELN, RFT1, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNU4-2, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUSC2, RYR2, SAMHD1, SARS, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO1, SCO2, SDHA, SEMA6B, SEPSECS, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGSH, SHQ1, SIK1, SIX3, SLC12A5, SLC13A3, SLC13A5, SLC16A2, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A22, SLC2A1, SLC31A1, SLC32A1, SLC35A1, SLC35A2, SLC35A3, SLC38A3, SLC39A8, SLC45A1, SLC4A10, SLC5A6, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMS, SNAP25, SNIP1, SNORD118, SNX27, SPATA5, SPATA5L1, SPR, SPTAN1, SPTBN1, SPTBN4, ST3GAL3, ST3GAL5, STAG1, STAMBP, STARD7, STRADA, STX1B, STXBP1, SUCLA2, SUCLG1, SUOX, SURF1, SYN1, SYNCRIP, SYNGAP1, SYNJ1, SZT2, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCK, TBL1XR1, TCF4, TDP2, TELO2, TET3, TFE3, TIAM1, TIMM50, TMEM106B, TMEM222, TMEM63B, TMEM70, TMX2, TNK2, TNPO2, TPP1, TRAF7, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIM8, TRIP13, TRIT1, TRPM3, TRPM6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TXNRD1, U2AF2, UBA5, UBAP2L, UBE2A, UBE3A, UBR7, UFC1, UFM1, UFSP2, UGDH, UGP2, UNC80, USP18, USP7, VAMP2, VARS, VLDLR, VPS11, VPS50, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WNK3, WWOX, XK, YIF1B,</p>			
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YIPF5, YWHAG, ZBTB18, ZDHC9, ZEB2, ZMIZ1, ZMYM2, ZNF142, ZNF335); SNV, CNV				
Familiärer Brust- und Eierstockkrebs (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, ATM, TP53, APC, ATRIP, BLM, CDKN1B, CDKN2A, CTNNA1, DICER1, EPCAM, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, LLGL2, MUTYH, NBN, NF1, NTHL1, POLD1, POLE, POLH, PTCH1, SLX4, SMARCA4, SMARCB1, SUFU, TRIM37, UBE2T, WRN, WT1); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_HBOC(Tier1)_v2026 und Tumordisp_HBOC(Tier2)_v2026, Pipeline V.2	25.07.2025
Familiärer Brust- und Eierstockkrebs (virtuelles Panel, Illumina PCR-Free Library Prep) (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, ATM, TP53, APC, ATRIP, BLM, CDKN1B, CDKN2A, CTNNA1, DICER1, EPCAM, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, LLGL2, MUTYH, NBN, NF1, NTHL1, POLD1, POLE, POLH, PTCH1, SLX4, SMARCA4, SMARCB1, SUFU, TRIM37, UBE2T, WRN, WT1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_HBOC(Tier1)_v2026 und Tumordisp_HBOC(Tier2)_v2026, Pipeline V.2	25.07.2025
Familiärer Brust- und Eierstockkrebs (Therapierelevanz) (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (BRCA1, BRCA2); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_BRCA1/2_only, Pipeline V.2	25.07.2025
Familiärer Brust- und Eierstockkrebs (Therapierelevanz) (virtuelles Panel, Illumina PCR-Free Library Prep) (BRCA1, BRCA2); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_BRCA1/2_only, Pipeline V.2	25.07.2025
Familiärer Brustkrebs (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ATM, ATRIP, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CDKN1B, CDKN2A, CHEK2, CTNNA1, EPCAM, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MLH1, MSH2, MSH6,	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_BC(Tier1)_v2026 und Tumordisp_BC(Tier2)_v2026, Pipeline V.2	25.07.2025

MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, POLH, PTEN, RAD51C, RAD51D, SLX4, STK11, TP53, UBE2T, WRN); SNV				
Familiärer Brustkrebs (virtuelles Panel, Illumina PCR-Free Library Prep) (ATM, ATRIP, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CDKN1B, CDKN2A, CHEK2, CTNNA1, EPCAM, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, POLH, PTEN, RAD51C, RAD51D, SLX4, STK11, TP53, UBE2T, WRN); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_BC(Tier1)_v2026 und Tumordisp_BC(Tier2)_v2026, Pipeline V.2	25.07.2025
Familiärer Eierstockkrebs (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, ATM, BLM, BRCA1, BRCA2, BRIP1, CDKN2A, CHEK2, DICER1, EPCAM, ERCC3, ERCC4, ERCC5, LLGL2, MLH1, MSH2, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTCH1, PTEN, RAD51C, RAD51D, SMARCA4, SMARCB1, STK11, SUFU, TP53, TRIM37, WT1); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_OC(Tier1)_v2026 und Tumordisp_OC(Tier2)_v2026, Pipeline V.2	25.07.2025
Familiärer Eierstockkrebs (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, ATM, BLM, BRCA1, BRCA2, BRIP1, CDKN2A, CHEK2, DICER1, EPCAM, ERCC3, ERCC4, ERCC5, LLGL2, MLH1, MSH2, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTCH1, PTEN, RAD51C, RAD51D, SMARCA4, SMARCB1, STK11, SUFU, TP53, TRIM37, WT1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_OC(Tier1)_v2026 und Tumordisp_OC(Tier2)_v2026, Pipeline V.2	25.07.2025
Fluoropyrimidin Toxizität (DPD-Gen: dbSNP rs3918290, rs55886062, rs67376798, rs56038477)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cyclor	GEN-AA-Genotypisierung LaCar V.2	15.11.2023
GIST (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, BMPR1A, CDH1, CTNNA1, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, NTHL1, PDGFRA, SDHA, SDHB, SDHC, SDHD); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_GIST_v2025_only1Tier, Pipeline V.2	25.07.2025

GIST (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, BMPR1A, CDH1, CTNNA1, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, NTHL1, PDGFRA, SDHA, SDHB, SDHC, SDHD); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_GIST_v2025_only1Tier, Pipeline V.2	25.07.2025
Hautkrebs/Melanom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ACD, BAP1, BRCA2, CDK4, CDKN2A, CYLD, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FLCN, GNAS, LEMD3, MC1R, MITF, POLH, POT1, PRDM10, PTCH1, PTEN, RB1, RECQL4, SUFU, TERF2IP, TERT, TP53, WRN, XPA, XPC); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Melanome_v.2025, Pipeline V.2	02.09.2025
Hautkrebs/Melanom (virtuelles Panel, Illumina PCR-Free Library Prep) (ACD, BAP1, BRCA2, CDK4, CDKN2A, CYLD, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FLCN, GNAS, LEMD3, MC1R, MITF, POLH, POT1, PRDM10, PTCH1, PTEN, RB1, RECQL4, SUFU, TERF2IP, TERT, TP53, WRN, XPA, XPC); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Melanome_v.2025, Pipeline V.2	02.09.2025
HCM (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ACTC1, ACTN2, ALPK3, ATAD3A, CACNA1C, CALR3, CAV3, COX15, CSRP3, DES, FHL1, FHOD3, FLNC, FXN, GAA, GLA, GYG1, JPH2, KLF10, KLHL24, LAMP2, MT-TI, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, RPS6KB1, SLC25A4, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, TULP3, VCL); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_HCM_v2026_Tier1 und Kardio_HCM_v2026_Tier2, Pipeline V.2	20.08.2025
HCM (virtuelles Panel, Illumina PCR-Free Library Prep) (ACTC1, ACTN2, ALPK3, ATAD3A, CACNA1C, CALR3, CAV3, COX15, CSRP3, DES, FHL1, FHOD3, FLNC, FXN, GAA, GLA, GYG1, JPH2, KLF10, KLHL24, LAMP2, MT-TI, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, RPS6KB1, SLC25A4, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, TULP3, VCL); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_HCM_v2026_Tier1 und Kardio_HCM_v2026_Tier2, Pipeline V.2	20.08.2025

<p>Hereditäre Neuropathien (virtuelles Panel, Illumina PCR-Free Library Prep) (AARS1, ABCA1, ABHD12, AGTPBP1, AGXT, AIFM1, AP1S1, APOA1, APTX, ARHGEF10, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CD59, CFAP276, CHCHD10, CNTNAP1, COA7, COX6A1, CPOX, CTD1P1, CYP27A1, DARS2, DCTN1, DEGS1, DNAJB2, DNAJC3, DNMT2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, ETFDH, FAH, FAM126A, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FXN, GALC, GAN, GART, GBA2, GDAP1, GJB1, GJC2, GLA, GNB4, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HSPB1, HSPB8, IARS2, IGHMBP2, INF2, JAG1, KCNA2, KIF1A, KIF5A, LITAF, LMNA, LRP12, LRSAM1, LYST, MCM3AP, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTPP, MYH14, NAGA, NDC1, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, OPA3, PDHA1, PDYN, PEX10, PEX7, PHYH, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, PPOX, PRDM12, PRKCG, PRNP, PRPS1, PRX, PTEN, PTPN11, PTRH2, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN9A, SCYL1, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SORD, SOX10, SPAST, SPG11, SPG7, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TFG, TRIM2, TRPA1, TRPV4, TTPA, TTR, TUBB3, TWNK, TYMP, VCP, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, XRCC1, YARS1, ZFYVE26); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina PCR-Free Library Prep V.3, HereditäreNeuropathien_v2025, Pipeline V.2</p>	<p>02.09.2025</p>
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Hereditäres Phäochromozytom-Paragangliom-Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (DLST, EGLN1, EPAS1, FH, MAX, MDH2, MEN1, MERTK, MET, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SUCLG2, TMEM127, VHL); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_PPGL_v2025, Pipeline V.2	25.07.2025
Hereditäres Phäochromozytom-Paragangliom-Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (DLST, EGLN1, EPAS1, FH, MAX, MDH2, MEN1, MERTK, MET, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SUCLG2, TMEM127, VHL); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_PPGL_v2025, Pipeline V.2	25.07.2025
Hirntumor (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, ATM, BRCA2, CDKN2A, ELP1, GPR161, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, POT1, PTEN, SMARCB1, SUFU, TP53, TSC1, TSC2); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Hirntumor_v.2025, Pipeline V.2	29.07.2025
Hirntumor (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, ATM, BRCA2, CDKN2A, ELP1, GPR161, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, POT1, PTEN, SMARCB1, SUFU, TP53, TSC1, TSC2); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Hirntumor_v.2025, Pipeline V.2	29.07.2025
Joubert-Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (AHI1, AIPL1, ARL13B, ARL6, ARMC9, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP290, CEP41, CPLANE1, CRB1, CRX, CSPP1, GUCY2D, HYLS1, 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); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Syndrom_Joubert_v2025, Pipeline V.2	10.03.2026

<p>Joubert-Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2CD3, CC2D2A, CCDC32, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP410, CILK1, CPLANE1, CRB2, CSPP1, DDX59, DHCR7, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, EXOC3L2, FAM149B1, GLI3, GLIS2, HNF1B, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT74, IFT80, IFT81, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF7, LAMA1, LZTFL1, MAPKBP1, MKKS, MKS1, NEK1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIBF1, PKD1, PKD2, PKHD1, PMM2, POC1B, RPGRI1L, SBDS, SCLT1, SDCCAG8, SUFU, TBC1D32, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TTC21B, TTC8, TXNDC15, VPS13B, WDPCP, WDR19, WDR35, XPNPEP3, ZNF423, ZSWIM6); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Joubert_v2026, Pipeline V.2</p>	<p>10.03.2026</p>
<p>Kardiomyopathien (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (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,</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>Sequence Capture, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina Exome 2.5 V.1, Kardio_Kardiomyopathien-breit_v2025_Tier1 und Kardio_Kardiomyopathien-breit_v2025_Tier2, Pipeline V.2</p>	<p>07.10.2025</p>

<p>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, 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, UQCC2, CTF1, DTNA, GATAD1, ILK, LAMA4, NEBL, PLEKHM2, PSEN1); SNV</p>				
<p>Kardiomyopathien (virtuelles Panel, Illumina PCR-Free Library Prep) (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,</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_Kardiomyopathien-breit_v2025_Tier1 und Kardio_Kardiomyopathien-breit_v2025_Tier2, Pipeline V.2</p>	<p>07.10.2025</p>

<p>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, 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, UQCC2, CTF1, DTNA, GATAD1, ILK, LAMA4, NEBL, PLEKHM2, PSEN1); SNV, CNV</p>				
<p>Leukodystrophie (virtuelles Panel, Illumina PCR-Free Library Prep) (ABCB7, ABCD1, ACSL4, AFF2, AIFM1, ALAS2, ALG13, AMER1, AP1S2, ARHGEF9, ARSE, ARX, ATP6AP1, ATP6AP2, ATP7A, ATRX, BCAP31, BCOR, BRWD3, CASK, CCDC22, CDK16, CDKL5, CLCN4, CNKSR2, COX7B, CUL4B, CXorf56, DCX, DDX3X, DKC1, DLG3, DMD, EBP, EIF2S3,</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina PCR-Free Library Prep V.3, Other_Leukodystrophy_v.2025_Tier1 und Other_Leukodystrophy_adultonset_v.2025_Tier2, Pipeline V.2</p>	<p>13.03.2026</p>

FAM50A, FGD1, FMR1, FRMPD4, FTSJ1, GDI1, GJB1, GK, GLA, GLRA2, GPC3, GPC4, GRIA3, HCCS, HCFC1, HDAC8, HNRNPH2, HPRT1, HSD17B10, HUWE1, IDS, IKBKG, IL1RAPL1, IQSEC2, KDM5C, KDM6A, KIF4A, L1CAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MSL3, NAA10, NDP, NDUFA1, NDUFB11, NEXMIF, NHS, NKAP, NLGN3, NONO, NSDHL, OCRL, OFD1, OGT, OPHN1, OTC, OTUD5, PAK3, PCDH19, PDHA1, PDK3, PGK1, PHF6, PHF8, PHKA1, PHKA2, PIGA, PLP1, POLA1, PORCN, PPP1R3F, PQBP1, PRPS1, PTCHD1, RAB39B, RBM10, RLIM, RNF113A, RPL10, RPS6KA3, SLC16A2, SLC35A2, SLC6A8, SLC9A6, SMARCA1, SMC1A, SMS, SRPK3, SSR4, STAG2, STS, SYN1, SYP, TAF1, TAZ, TCEAL1, TFE3, THOC2, TIMM8A, UBE2A, UPF3B, USP9X, WDR45, WNK3, ZC4H2, ZDHHC9, ZFX, ZMYM3, ZNF711); SNV, CNV				
Li-Fraumeni-Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, BRCA1, BRCA2, CDH1, CHEK2, GNAS, GPC3, GPR161, HRAS, KRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NRAS, NTHL1, PALB2, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD51C, RAD51D, RAF1, RB1, RECQL4, REST, RET, RIT1, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, STK11, SUFU, TERC, TERT, TMEM127, TP53, TRIM37, TSC1, TSC2, VHL, WT1, XRCC2); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_LiFraumeni_v2025, Pipeline V.2	11.07.2025
Li-Fraumeni-Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, BRCA1, BRCA2, CDH1, CHEK2, GNAS, GPC3, GPR161, HRAS, KRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NRAS, NTHL1, PALB2, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD51C, RAD51D, RAF1, RB1,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_LiFraumeni_v2025, Pipeline V.2	11.07.2025

RECQL4, REST, RET, RIT1, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, STK11, SUFU, TERC, TERT, TMEM127, TP53, TRIM37, TSC1, TSC2, VHL, WT1, XRCC2); SNV, CNV				
Long_QT (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TECRL, TRDN); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_LongQT_v2025_Tier1 und Kardio_LongQT_v2025_Tier2, Pipeline V.2	15.08.2025
Long_QT (virtuelles Panel, Illumina PCR-Free Library Prep) (ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TECRL, TRDN); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_LongQT_v2025_Tier1 und Kardio_LongQT_v2025_Tier2, Pipeline V.2	15.08.2025
Lynch-Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (MLH1, PMS2, MSH6, MSH2, EPCAM); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Lynch-syndrom_v2025, Pipeline V.2	14.07.2025
Lynch-Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (MLH1, PMS2, MSH6, MSH2, EPCAM); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Lynch-syndrom_v2025, Pipeline V.2	14.07.2025
Magenkrebs (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, BMPR1A, CDH1, CHEK2, CTNNA1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PDGFRA, PMS2, POLD1, POLE, PTPN11, SMAD4, STK11, TP53); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Magenkrebs-breit_v2025_only1Tier, Pipeline V.2	01.08.2025
Magenkrebs (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, BMPR1A, CDH1, CHEK2, CTNNA1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PDGFRA, PMS2, POLD1, POLE, PTPN11, SMAD4, STK11, TP53); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Magenkrebs-breit_v2025_only1Tier, Pipeline V.2	01.08.2025
Marfan-Syndrom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (FBN1, TGFBR1, TGFBR2, IPO8, COL1A2, FKBP14); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Syndrom_marfan_v2025, Pipeline V.2	26.03.2026
Marfan-Syndrom (virtuelles Panel, Illumina PCR-Free Library Prep) (FBN1, TGFBR1, TGFBR2, IPO8, COL1A2, FKBP14); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_marfan_v2025, Pipeline V.2	26.03.2026

Mavacamten Therapie (CYP2C19-Gen: CYP2C19*2, CYP2C19*3)	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Pipeline V.2	15.11.2023
Mavacamten Therapie (CYP2C19-Gen: CYP2C19*2, CYP2C19*3)	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Pipeline V.2	15.11.2023
Medulloblastom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, BRCA2, DICER1, ELP1, GPC3, GRP161, NBN, PALB2, PTCH1, RET, SEC23B, SUFU, TP53); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Medulloblastom_v.2026_only1Tier, Pipeline V.2	10.03.2026
Medulloblastom (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, BRCA2, DICER1, ELP1, GPC3, GRP161, NBN, PALB2, PTCH1, RET, SEC23B, SUFU, TP53); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Medulloblastom_v.2026_only1Tier, Pipeline V.2	10.03.2026
MODY (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ABCC8, APPL1, BLK, BSCL2, CEL, CNOT1, EIF2AK3, EIF2B1, EIF2S3, FICD, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, KCNJ11, KLF11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, ONECUT1, PAX1, PDIA6, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57, ZNF808); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Other_MODY_v.2025, Pipeline V.2	26.09.2025
MODY (virtuelles Panel, Illumina PCR-Free Library Prep) (ABCC8, APPL1, BLK, BSCL2, CEL, CNOT1, EIF2AK3, EIF2B1, EIF2S3, FICD, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, KCNJ11, KLF11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, ONECUT1, PAX1, PDIA6, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57, ZNF808); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Other_MODY_v.2025, Pipeline V.2	26.09.2025
Morbus Bechterew (HLA-B27 Subtypen B*2702 und B*2705)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler	GEN-AA-Genotypisierung LaCar V.2	15.11.2023

Morbus Bechterew (HLA-B27 Subtypen B*2702, B*2703, B*2704 und B*2705)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler	GEN-AA-Genotypisierung-HLA-B27-direct LaCar V.2	15.11.2023
Morbus Stargardt (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ABCA4, ARMS2, BEST1, C1QTNF5, CDH3, CFH, CLN3, CNGB3, CRX, CTNNA1, DRAM2, ELOVL4, GUCA1A, IMPG1, IMPG2, IRX1, MFSD8, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTL5); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Sensorineural_Stargardt_v2025_Tier1+2, Pipeline V.2	31.03.2026
Morbus Stargardt (virtuelles Panel, Illumina PCR-Free Library Prep) (ABCA4, ARMS2, BEST1, C1QTNF5, CDH3, CFH, CLN3, CNGB3, CRX, CTNNA1, DRAM2, ELOVL4, GUCA1A, IMPG1, IMPG2, IRX1, MFSD8, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTL5); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensorineural_Stargardt_v2025_Tier1+2, Pipeline V.2	31.03.2026
Neurofibromatose / Schwannomatose (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (BRAF, BRCA2, BRIP1, EPCAM, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GNAS, HOXB13, KITLG, LZTR1, MAP2K1, MLH1, MSH2, MSH6, NF1, NF2, PALB2, PDGFRB, PMS2, POLA1, PRKAR1A, PTPN11, RAD51C, RAF1, RIT1, SLX4, SMARCB1, SOS1, SPRED1); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Neurofibromatosis_v.2025_Tier1 und Tumordisp_Neurofibromatosis_v.2025_Tier2, Pipeline V.2	01.08.2025
Neurofibromatose / Schwannomatose (virtuelles Panel, Illumina PCR-Free Library Prep) (BRAF, BRCA2, BRIP1, EPCAM, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GNAS, HOXB13, KITLG, LZTR1, MAP2K1, MLH1, MSH2, MSH6, NF1, NF2, PALB2, PDGFRB, PMS2, POLA1, PRKAR1A, PTPN11, RAD51C, RAF1, RIT1, SLX4, SMARCB1, SOS1, SPRED1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Neurofibromatosis_v.2025_Tier1 und Tumordisp_Neurofibromatosis_v.2025_Tier2, Pipeline V.2	01.08.2025

<p>Neuropathien (virtuelles Panel, Illumina PCR-Free Library Prep) (AARS1, ABCA1, ABHD12, AGTPBP1, AGXT, AIFM1, AP1S1, APOA1, APTX, ARHGEF10, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CD59, CFAP276, CHCHD10, CNTNAP1, COA7, COX6A1, CPOX, CTDP1, CYP27A1, DARS2, DCTN1, DEGS1, DNAJB2, DNAJC3, DNMT2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, ETFDH, FAH, FAM126A, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FXN, GALC, GAN, GART, GBA2, GDAP1, GJB1, GJC2, GLA, GNB4, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HSPB1, HSPB8, IARS2, IGHMBP2, INF2, JAG1, KCNA2, KIF1A, KIF5A, LITAF, LMNA, LRP12, LRSAM1, LYST, MCM3AP, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTPP, MYH14, NAGA, NDC1, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, OPA3, PDHA1, PDYN, PEX10, PEX7, PHYH, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, PPOX, PRDM12, PRKCG, PRNP, PRPS1, PRX, PTEN, PTPN11, PTRH2, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN9A, SCYL1, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SORD, SOX10, SPAST, SPG11, SPG7, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TFG, TRIM2, TRPA1, TRPV4, TTPA, TTR, TUBB3, TWNK, TYMP, VCP, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, XRCC1, YARS1, ZFYVE26); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina PCR-Free Library Prep V.3, Other_Neuropathien_v2025_Tier1+2, Pipeline V.2</p>	<p>02.09.2025</p>
<p>Nierenkarzinom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (BAP1, CDC73, CDKN1B, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, MET, MLH1, MSH2, MSH6, PMS2, PTEN, REST, SDHA, SDHAF2, SDHB, SDHC, SDHD, STK11, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1); SNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>Sequence Capture, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Nierenkarzinom_v.2025, Pipeline V.2</p>	<p>02.09.2025</p>

Nierenkarzinom (virtuelles Panel, Illumina PCR-Free Library Prep) (BAP1, CDC73, CDKN1B, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, MET, MLH1, MSH2, MSH6, PMS2, PTEN, REST, SDHA, SDHAF2, SDHB, SDHC, SDHD, STK11, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Nierenkarzinom_v.2025, Pipeline V.2	02.09.2025
Noonan Syndrom/RASopathie (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ARID2, BRAF, CBL, HRAS, ISCA-37431-Loss, KRAS, LZTR1, MAP2K1, MAP2K2, MAP4K4, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, RREB1, SHOC2, SOS1, SOS2, SPRED1, SPRED2); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Syndrom_Noonan_v2026_Tier1+2, Pipeline V.2	12.01.2026
Noonan Syndrom/RASopathie (virtuelles Panel, Illumina PCR-Free Library Prep) (ARID2, BRAF, CBL, HRAS, ISCA-37431-Loss, KRAS, LZTR1, MAP2K1, MAP2K2, MAP4K4, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, RREB1, SHOC2, SOS1, SOS2, SPRED1, SPRED2); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Noonan_v2026_Tier1+2, Pipeline V.2	12.01.2026
Optikusatrophie (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ACO2, AFG3L2, ALPK1, AP3B2, ATAD3A, ATG7, ATP1A3, BLOC1S1, BORCS8, BTD, C19orf12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, HIKESHI, HK1, HSD17B10, ISCA2, LETM1, LHX2, LRRC8C, MAG, MCAT, MECR, MFF, MFN2, MIEF1, MT-ATP6, MT-ND1, MT-ND4, MT-ND6, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDSS1, PDXK, POLG, RTN4IP1, SLC25A46, SLC44A1, SLC52A2, SNF8, SPG7, SSBP1, TFG, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Sensineural_Optic Atrophy_v2025_Tier1 und Sensineural_Optic Atrophy_v2025_Tier2, Pipeline V.2	16.05.2025
Optikusatrophie (virtuelles Panel, Illumina PCR-Free Library Prep) (ACO2, AFG3L2, ALPK1, AP3B2, ATAD3A, ATG7, ATP1A3, BLOC1S1, BORCS8, BTD, C19orf12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, HIKESHI, HK1, HSD17B10, ISCA2, LETM1, LHX2, LRRC8C, MAG, MCAT, MECR, MFF, MFN2, MIEF1, MT-ATP6, MT-ND1, MT-ND4, MT-ND6, MTRFR, NBAS, NDUFA12, NR2F1,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensineural_Optic Atrophy_v2025_Tier1 und Sensineural_Optic Atrophy_v2025_Tier2, Pipeline V.2	16.05.2025

OPA1, OPA3, PDSS1, PDXK, POLG, RTN4IP1, SLC25A46, SLC44A1, SLC52A2, SNF8, SPG7, SSBP1, TFG, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1); SNV, CNV				
PAH (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, GDF2, GGCX, KCNK3, KDR, NFU1, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4, TET2); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_PAH_v2025_Tier1 und Kardio_PAH_v2025_Tier2, Pipeline V.2	18.08.2025
PAH (virtuelles Panel, Illumina PCR-Free Library Prep) (ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, GDF2, GGCX, KCNK3, KDR, NFU1, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4, TET2); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_PAH_v2025_Tier1 und Kardio_PAH_v2025_Tier2, Pipeline V.2	18.08.2025
Pankreaskarzinom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, ATM, BMPR1A, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, PTEN, SMAD4, SPINK1, STK11, TP53, VHL); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_PankreasCA_v.2025Tier1+Tier2, Pipeline V.2	10.07.2025
Pankreaskarzinom (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, ATM, BMPR1A, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, PTEN, SMAD4, SPINK1, STK11, TP53, VHL); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_PankreasCA_v.2025Tier1+Tier2, Pipeline V.2	10.07.2025
Plötzlicher Herztod (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (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, MT-TI, MYBPC3, MYH6, MYH7,	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_plötzlicherHerztod_v.2025_Tier 1 und Tier 2, Pipeline V.2	18.03.2026

MYL2, MYL3, MYLK2, MYLK3, MYPN, MYZAP, NEXN, NKX2-5, NRAP, PHOX2B, PKP2, PLN, PPA2, PRDM16, PRKAG2, PRKAR1A, 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); SNV				
Plötzlicher Herztod (virtuelles Panel, Illumina PCR-Free Library Prep) (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, MT-TI, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYLK3, MYPN, MYZAP, NEXN, NKX2-5, NRAP, PHOX2B, PKP2, PLN, PPA2, PRDM16, PRKAG2, PRKAR1A, 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); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_plötzlicherHerztod_v.2025_Tier 1 und Tier 2, Pipeline V.2	18.03.2026
Polyposis (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, AXIN2, BMPR1A, EPCAM, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SLX4, SMAD4, STK11, UBE2T, XRCC2); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Polyposis_v.2025_Tier 1 und Tumordisp_Polyposis_v.2025_Tier 2, Pipeline V.2	28.07.2025

Polyposis (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, AXIN2, BMPR1A, EPCAM, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SLX4, SMAD4, STK11, UBE2T, XRCC2); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Polyposis_v.2025_Tier 1 und Tumordisp_Polyposis_v.2025_Tier 2, Pipeline V.2	28.07.2025
Polyzystische Nierenerkrankung (virtuelles Panel, Illumina PCR-Free Library Prep) (ALG5, ALG8, ALG9, ANKS6, CEP164, CEP290, CEP83, CFAP47, CLCN5, COL4A1, COL4A3, COL4A4, CYP24A1, CYS1, DNAJB11, DZIP1L, FLCN, GANAB, GLA, HNF1B, IFT140, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PRKCSH, SEC61A1, SEC63, TMEM67, TSC1, TSC2, TTC21B, TULP3, UMOD, VHL, WDR19, XPNPEP3, ZNF423); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Polyzystische Nierenerkrankung_v2026Tier1+2, Pipeline V.2	12.03.2026
Polyzystische Nierenerkrankung (virtuelles Panel, Illumina PCR-Free Library Prep) (ALG5, ALG8, ALG9, ANKS6, CEP164, CEP290, CEP83, CFAP47, CLCN5, COL4A1, COL4A3, COL4A4, CYP24A1, CYS1, DNAJB11, DZIP1L, FLCN, GANAB, GLA, HNF1B, IFT140, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PRKCSH, SEC61A1, SEC63, TMEM67, TSC1, TSC2, TTC21B, TULP3, UMOD, VHL, WDR19, XPNPEP3, ZNF423); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Polyzystische Nierenerkrankung_v2026Tier1+2, Pipeline V.2	12.03.2026
Prostatakrebs (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ATM, BRCA1, BRCA2, CDKN1B, CHEK2, EPCAM, GNAS, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51D, TP53): SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_ProstataCA_v.2025, Pipeline V.2	08.07.2025
Prostatakrebs (virtuelles Panel, Illumina PCR-Free Library Prep) (ATM, BRCA1, BRCA2, CDKN1B, CHEK2, EPCAM, GNAS, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51D, TP53): SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_ProstataCA_v.2025, Pipeline V.2	08.07.2025

<p>Retinitis Pigmentosa (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (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, AHR, CFAP20, DHX38, DYNC2H1, KIF3B, MVK, SEMA4A, SLC37A3, UBAP1L, VWA8, ADGRV1, BBS1, CFAP418, EXOSC2, ZNF408, PCARE, PRPH2, RCBTB1); SNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>Sequence Capture, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina Exome 2.5 V.1, Sensorineural_Retinitis_pigmentosa_v2026_Tier1 und Sensorineural_Retinitis_pigmentosa_v2026_Tier2, Pipeline V.2</p>	<p>26.03.2026</p>
<p>Retinitis Pigmentosa (virtuelles Panel, Illumina PCR-Free Library Prep) (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, AHR, CFAP20, DHX38, DYNC2H1, KIF3B, MVK, SEMA4A, SLC37A3, UBAP1L, VWA8, ADGRV1, BBS1, CFAP418, EXOSC2, ZNF408, PCARE, PRPH2, RCBTB1); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN</p>	<p>GEN-AA-Illumina PCR-Free Library Prep V.3, Sensorineural_Retinitis_pigmentosa_v2026_Tier1 und Tier2, Pipeline V.2</p>	<p>26.03.2026</p>

Rhabdomyolyse (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ABHD5, ACAD9, ACADM, ACADVL, AGL, ALDOA, AMACR, AMPD1, ANO5, ATP2A2, CACNA1S, CAV3, CHKB, COQ4, COQ8A, CPT1A, CPT2, DGUOK, DMD, DYSF, ENO3, ETFA, ETFB, ETFDH, FDX2, FKRP, FLAD1, GAA, GBE1, GMPPB, GYG1, GYS1, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, MLIP, MT-CO1, MT-CO2, MYH1, OBSCN, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, POLG, POLG2, PRKAG2, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SIL1, SLC22A5, SUCLA2, TAMM41, TANGO2, TK2, TSFM, TYMP); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Other_Rhabdomyolysis_v.2026, Pipeline V.2	21.01.2026
Rhabdomyolyse (virtuelles Panel, Illumina PCR-Free Library Prep) (ABHD5, ACAD9, ACADM, ACADVL, AGL, ALDOA, AMACR, AMPD1, ANO5, ATP2A2, CACNA1S, CAV3, CHKB, COQ4, COQ8A, CPT1A, CPT2, DGUOK, DMD, DYSF, ENO3, ETFA, ETFB, ETFDH, FDX2, FKRP, FLAD1, GAA, GBE1, GMPPB, GYG1, GYS1, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, MLIP, MT-CO1, MT-CO2, MYH1, OBSCN, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, POLG, POLG2, PRKAG2, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SIL1, SLC22A5, SUCLA2, TAMM41, TANGO2, TK2, TSFM, TYMP); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Other_Rhabdomyolysis_v.2026, Pipeline V.2	21.01.2026
Sarkoma (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (MTAP, PDGFRA, RECQL4, SQSTM1, TBXT, TP53, EXT1, EXT2, NF1, APC, ATM, ATR, BLM, BRCA2, BUB1B, CDKN1C, ERCC2, FANCC, FH, HRAS, KIT, MLH1, MSH2, MSH6, NBN, PMS2, RB1, SDHA, SDHB, SDHD, SMARCA4, WRN); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Sarcoma_v.2025_Tier1 und Tumordisp_Sarcoma_v.2025_Tier2, Pipeline V.2	27.03.2026
Sarkoma (virtuelles Panel, Illumina PCR-Free Library Prep) (MTAP, PDGFRA, RECQL4, SQSTM1, TBXT, TP53, EXT1, EXT2, NF1, APC, ATM, ATR, BLM, BRCA2, BUB1B, CDKN1C, ERCC2, FANCC, FH, HRAS, KIT, MLH1, MSH2, MSH6, NBN, PMS2, RB1, SDHA, SDHB, SDHD, SMARCA4, WRN); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Sarcoma_v.2025_Tier1 und Tumordisp_Sarcoma_v.2025_Tier2, Pipeline V.2	27.03.2026

Schilddrüsenkarzinom (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (APC, CDC73, CDKN1B, CHEK2, DICER1, FLCN, FOXE1, GNAS, MSH2, MUTYH, PRKAR1A, PTEN, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SLC5A5, TP53, TSC1, TSC2, WRN); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_Schilddrüsen CA_v2025_Tier1 und Tumordisp_Schilddrüsen CA_v2025_Tier2, Pipeline V.2	25.07.2025
Schilddrüsenkarzinom (virtuelles Panel, Illumina PCR-Free Library Prep) (APC, CDC73, CDKN1B, CHEK2, DICER1, FLCN, FOXE1, GNAS, MSH2, MUTYH, PRKAR1A, PTEN, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SLC5A5, TP53, TSC1, TSC2, WRN); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Schilddrüsen CA_v2025_Tier1 und Tumordisp_Schilddrüsen CA_v2025_Tier2, Pipeline V.2	25.07.2025
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; DNA	PCR und Sanger-Sequenzierung	GEN-AA-Sanger-Sequenzierung V.3	15.11.2023
Sensorineurale Schwerhörigkeit (virtuelles Panel, Illumina PCR-Free Library Prep) (ABCC1, ABHD12, ACOX1, ACTG1, ADGRV1, AFG2A, AIFM1, ALMS1, AP1S1, ATP11A, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BMP4, BSND, CABP2, CCDC50, CDC14A, CDC6, CDH23, CDT1, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, CLRN2, COCH, COG4, COL11A1, COL11A2, COL2A1, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRLS1, CRYM, DAP3, DFNA5, DFN59, DHODH, DIABLO, DIAPH1, DIAPH3, DMXL2, DNAJC3, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, EIF4A3, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EXOSC2, EYA1, EYA4, FDXR, FGF10, FGF3, FGFR2, FGFR3, FOXF2, FOXI3, FRAS1, FREM2, GATA3, GDF6, GGPS1, GIPC3, GJB2, GJB3, GJB6, GNAI3, GPR156, GPSM2, GREB1L, GRHL2, GRIP1, GRXCR1, GRXCR2, GSC, GSDME, HAAO, HARS2, HGF, HMX1, HOMER2, HOXA2, HSD17B4, HSPA9, IARS2, ILDR1, KARS, KARS1, KCNE1, KCNJ10, KCNJ16, KCNQ1, KCNQ4, KDM3B, KDM6A, KIAA1024L,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensineural_Hearingloss_v2025_Tier1 und Sensineural_Hearingloss_v2025_Tier2, Pipeline V.2	29.04.2025

KIT, KITLG, KMT2D, LARS2, LETM1, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MASP1, MCM2, MET, MIR96, MITF, MN1, MORC2, MPZL2, MSRB3, MT-RNR1, MT-TS1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP12, OFD1, OGDHL, OPA1, ORC1, ORC4, ORC6, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTX2, OXR1, P2RX2, PAX2, PAX3, PBX1, PCDH15, PDSS1, PDZD7, PHYH, PJVK, PKHD1L1, PLCB4, PLS1, PLXNB2, PMP22, PNPT1, POLR1A, POLR1C, POLR1D, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RFC4, RIPOR2, RNF220, ROR1, RPS28, S1PR2, SALL1, SALL4, SCD5, SERAC1, SERPINB6, SF3B4, SGPL1, SIX1, SIX5, SLC12A2, SLC17A8, SLC26A4, SLC26A5, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, SOX2, SPATA5, SPATA5L1, SPATC1L, SPNS2, SPTBN4, STRC, STX4, STXBP3, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, THOC1, TIMM8A, TMC1, TMIE, TMPRSS3, TMTC2, TNC, TOP2B, TPRN, TRIOBP, TUBB4B, TWNK, USH1C, USH1G, USH2A, USP48, WBP2, WFS1, WHRN, YARS); SNV, CNV				
Sensorineurale Schwerhörigkeit (<i>GJB2</i>)	EDTA-Blut, DNA; DNA	PCR und Sanger-Sequenzierung	GEN-AA-Sanger-Sequenzierung V.3	15.11.2023
Short_QT (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCN5A, SLC4A3); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_ShortQT_v2025_Tier1only, Pipeline V.2	08.08.2025
Short_QT (virtuelles Panel, Illumina PCR-Free Library Prep) (CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCN5A, SLC4A3); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_ShortQT_v2025_Tier1only, Pipeline V.2	08.08.2025
Solide kindliche Tumore (virtuelles Panel, Illumina PCR-Free Library Prep) (ACD, AKT1, ALK, ANKRD26, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTC1, CTR9, DDB2, DDX41, DICER1, DIS3L2, DKC1, DNAJC21,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Kindliche-Tumore_v2025_Tier1 und Tumordisp_Kindliche-Tumore_v2025_Tier2, Pipeline V.2	21.07.2025

ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAS, FH, GATA1, GATA2, GBA, GNAS, GPC3, GPR161, HAX1, HRAS, IKZF1, ITK, KIT, KRAS, LIG4, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAX, MEN1, MET, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NHP2, NOP10, NRAS, NSD1, NTHL1, NYNRIN, PALB2, PARN, PAX5, PDGFRA, PDGFRB, PHOX2B, PMS2, POLD1, POLE, POLH, POT1, PPP1CB, PRKAR1A, PTCH1, PTEN, PTPN11, RAD51, RAD51C, RAD51D, RAF1, RB1, RECQL4, REST, RET, RIT1, RPL11, RPL15, RPL23, RPL26, RPL31, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, RUNX1, SBDS, SEC23B, SERPINA1, SH2D1A, SHOC2, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SPRED1, STAT3, STK11, SUFU, TERC, TERT, TINF2, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRN, WT1, XPA, XPC, XRCC2); SNV, CNV				
Stäbchen-Zapfen Dystrophie (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (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); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Sensorineural_Zapfen-Stäbchen-Dystrophie_v2025_only1Tier, Pipeline V.2	01.04.2026
Stäbchen-Zapfen Dystrophie (virtuelles Panel, Illumina PCR-Free Library Prep) (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); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensorineural_Zapfen-Stäbchen-Dystrophie_v2025_only1Tier, Pipeline V.2	01.04.2026

TAAD (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (ABL1, ACTA2, ADAMTSL4, ARIH1, ASPH, BGN, CBS, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, EMILIN1, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FOXE3, HCN4, IPO8, JAG1, LMOD1, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, NPR3, PLOD1, PMEPA1, PRKG1, ROBO4, SECISBP2, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TBX20, TGFB2, TGFB3, TGFB1, TGFB2, THSD4); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina Exome 2.5 V.1, Kardio_TAAD_v2026 Tier 1 und Kardio_TAAD_v2026 Tier 2, Pipeline V.2	16.01.2026
TAAD (virtuelles Panel, Illumina PCR-Free Library Prep) (ABL1, ACTA2, ADAMTSL4, ARIH1, ASPH, BGN, CBS, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, EMILIN1, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FOXE3, HCN4, IPO8, JAG1, LMOD1, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, NPR3, PLOD1, PMEPA1, PRKG1, ROBO4, SECISBP2, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TBX20, TGFB2, TGFB3, TGFB1, TGFB2, THSD4); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_TAAD_v2026 Tier 1 und Kardio_TAAD_v2026 Tier 2, Pipeline V.2	16.01.2026
Thrombophilie (F5-Gen: dbSNP rs6025, F2-Gen: rs1799963)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler	GEN-AA-Genotypisierung LaCar V.2	15.11.2023
Tumordisposition breit (virtuelles Panel, Illumina PCR-Free Library Prep) (ABL1, ABRAXAS1, ACD, ACVR1, ACVR1B, AIP, AKT1, ALK, ALOX12B, ANKRD26, APC, ASXL1, ATM, ATR, ATRX, AURKA, AURKB, AXIN1, AXIN2, BAP1, BARD1, BBC3, BCL10, BCL6, BCR, BLM, BMP1R1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1, BUB1B, CALR, CARD11, CASP8, CBL, CCND1, CCND2, CCND3, CCNE1, CDC73, CDH1, CDK12, CDK4, CDKN1A, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CDKN2C, CEBPA, CEP57, CHEK2, CRKL, CRLF2, CSF1R, CSF3R, CTLA4, CTNNA1, CTNNA1, CTNNB1, CTR9, CTCR, CUX1, CXCR4, CYLD, DCUN1D1, DDB2, DDR2, DDX41, DICER1,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_breit_Tier1+2_v2026, Pipeline V.2	09.01.2026

DIS3L2, DKC1, DLST, DNAJB1, DNAJC21, DNMT3A, DNMT3B, EGFR, EGLN1, ELP1, EPAS1, EPCAM, ERBB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ESR1, ETV6, EWSR1, EXT1, EXT2, EZH2, FAM123B, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FAT1, FGFR1, FGFR2, FGFR3, FGFR4, FH, FLCN, FLT3, FLT4, FOXA1, FOXE1, FOXO1, FRS2, FYN, GALNT12, GATA1, GATA2, GATA3, GATA4, GATA6, GBA, GNAS, GPC3, GPR101, GPR124, GPR161, GPS2, GREM1, GRM3, HABP2, HNF1A, HOXB13, HRAS, ID3, IDH1, IDH2, IKZF1, ITK, JAK2, JUN, KDR, KIF1B, KIT, KMT2D, KRAS, LATS1, LATS2, LIG4, LMO1, LRP1B, LYN, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAP2K4, MAX, MBD4, MC1R, MCL1, MDC1, MDH2, MDM2, MDM4, MEF2B, MEN1, MET, MITF, MLH1, MLH3, MPL, MRE11, MRE11A, MSH2, MSH3, MSH6, MST1R, MTOR, MUTYH, MYB, MYC, MYCL1, MYCN, MYD88, NAF1, NBN, NEGR1, NF1, NF2, NHP2, NKX2-1, NKX3-1, NOP10, NOTCH1, NPM1, NRAS, NSD1, NTHL1, NUTM1, PALB2, PARN, PARP1, PAX3, PAX5, PAX7, PBRM1, PDCD1, PDGFRA, PDGFRB, PHOX2B, PIK3CA, PIM1, PMS2, POLD1, POLE, POLH, POT1, PPM1D, PREX2, PRF1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, PTPN11, PTPRT, RAB35, RABL3, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD54L, RAF1, RARA, RASA1, RB1, RECQL4, REL, REST, RET, RIT1, RNF43, RPA1, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SERPINA1, SETBP1, SETD2, SF3B1, SFTPA1, SH2B3, SH2D1A, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SMO, SOS1, SOX9, SPINK1, SPRED1, SRC, SRGAP1, SRP72, SRSF2, STAT4, STK11, STN1, SUFU, SYK, TCF3, TERC, TERF2IP, TERT, TET2, TFE3, TGFBR1, TGFBR2, TINF2, TMEM127, TNFAIP3, TNFRSF14, TOP1, TOP2A, TP53, TRAF7, TRIM28,				
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TRIM37, TRIP13, TSC1, TSC2, TSHR, U2AF1, UBE2T, VEGFA, VHL, WRAP53, WRN, WT1, XIAP, XPA, XPC, XRCC2, ZFH3, ZNF217); SNV, CNV				
Ungerichtete Auswertung von Kopienzahlveränderungen > 50kB (Illumina PCR-Free Library Prep)	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Pipeline V.2	15.09.2025
Unklare Erkrankungen mit wahrscheinlich erblicher Ursache und Auswertung eines Morbid Genes Panels; SNV, CNV (Illumina PCR-Free Library Prep)	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, MorbidGenes Panel V.2026_02, Pipeline V.2	17.03.2026
Unklare Erkrankungen mit wahrscheinlich erblicher Ursache und Auswertung von Varianten mit HPO Terms; SNV, CNV (Illumina PCR-Free Library Prep)	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Pipeline V.2	15.11.2023
Ziliopathie (virtuelles Panel, Illumina PCR-Free Library Prep) (ADAMTS9, AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BRWD1, C2CD3, CBY1, CC2D2A, CCDC103, CCDC32, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP55, CEP83, CFAP221, CFAP298, CFAP300, CFAP410, CFAP418, CFAP74, CFTR, CILK1, CPLANE1, CRB2, CSPP1, CYS1, DAW1, DCDC2, DDX59, DHCR7, DLG5, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2I2, DYNLT2B, EVC, EVC2, EXOC3L2, FAM149B1, FOXJ1, GAS2L2, GAS8, GLI3, GLIS2, GRK2, HNF1B, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, IQCE, KATNIP, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, LAMA1, LBR, LRRC45, LRRC56, LZTFL1, MAPKBP1, MCIDAS, MDM1, MKKS,	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, DRAGEN	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Ziliopathie_v2026Tier1+2, Pipeline V.2	16.03.2026

MKS1, NEK1, NEK10, NEK8, NME5, NME8, NPHP1, NPHP3, NPHP4, ODAD1, ODAD2, ODAD3, OFD1, PDIA6, PIBF1, PIK3C2A, PKD1, PKD2, PKHD1, PMM2, POC1B, POC5, PRKACA, PRKACB, PSKH1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SBDS, SCLT1, SDCCAG8, SPAG1, STK36, SUFU, TBC1D32, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TMEM72, TOGARAM1, TRAF3IP1, TTC12, TTC21B, TTC8, TXNDC15, VPS13B, WDPCP, WDR19, WDR35, XPNPEP3, ZMYND10, ZNF423, ZSWIM6); SNV, CNV				
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