Liste der im flexiblen Bereich akkreditierten Verfahren

Stand: 17.10.2025

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	aufgenommen am
Unklare Erkrankungen mit wahrscheinlich erblicher Ursache und Auswertung von Varianten mit HPO Terms; SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Pipeline V.2	15.11.2023
Fluoropyrimidin Toxizität (DPD-Gen: dbSNP rs3918290, rs55886062, rs67376798, rs56038477)	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 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
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

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Hämochromatose (HFE-Gen: rs1800562, rs1799945)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler	GEN-AA-HFE Genotypisierung V.1	15.11.2023
Siponimod Toxizität (CYP2C9-Gen: CYP2C9*2 (rs1799853) und CYP2C9*3 (rs1057910))	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler	GEN-AA-CYP2C9 und CYP2C19-Genotypisierung LaCar V.1	24.11.2024
Mavacamten Toxizität (CYP2C19-Gen: CYP2C19*2, CYP2C19*3 und CYP2C19*17)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler	GEN-AA-CYP2C9 und CYP2C19-Genotypisierung LaCar V.1	24.11.2024
Noonan Syndrom/RASopathie (*BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Noonan_v2024_Tier1+2, Pipeline V.2	30.05.2024
Aicardi-Goutières-Syndrom (*ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Aicardi-Goutières_v2023, Pipeline V.2	30.05.2024
Bardet-Biedl-Syndrom (*ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TTC8, WDPCP*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Bardet-Biedl_v2023, Pipeline V.2	30.05.2024
CHARGE-Syndrom (*CHD7*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_CHARGE_v2023, Pipeline V.2	30.05.2024
Cornelia-de-Lange-Syndrom (*HDAC8, NIPBL, RAD21, SMC3*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Cornelia-de-Lange_v2023, Pipeline V.2	30.05.2024
Ehlers-Danlos Syndrom (*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, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Ehlers-Danlos_v.2024 T1+2 Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Coffin-Siris-Syndrom *(ARID1A, ARID1B, BICRA, HDAC8, NIPBL, PHF6, PIGV, RAD21, SMARCA2, SMARCA4, SMC3, TBC1D24*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Coffin-Siris_v2023, Pipeline V.2	30.05.2024
Kabuki-Syndrom (*CHD7, EYA1, FLNB, IRF6, KDM6A, KMT2D, SIX1, SIX5*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Kabuki_v2023, Pipeline V.2	30.05.2024
Rubenstein-Taybi-Syndrom (*CREBBP, EP300, FGFR1, FGFR2, GLI3, HOXD13, SRCAP, TWIST1*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Rubenstein_Taybi_v2023, Pipeline V.2	30.05.2024
Smith-Lemli-Opitz-Syndrom (*B9D2, BRAF, CBL, CC2D2A, CEP290, DHCR24, DHCR7, EBP, FDFT1, 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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Smith-Lemli-Opitz_v2023, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Epilepsie (*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, GABRD, GABRG2, GNAO1, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, 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, 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, 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, COL4A3BP, COG4, COG8, COL18A1, COL4A1, COL4A2, CPA6, CREBBP, CTU2, CUL3, CUX1, CUX2, DEAF1, DHX16, DLL1, DNAJC5, DNM1L, DYNC1H1, DYRK1A, EFHC1, EFTUD2, EHMT1, EIF2AK2, EIF4A2, EXT2, FAR1, FBXO11, FGFR3, FH, FOXG1, FRMD5, FZR1, GABRB1, 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, KDM6B, KIF1A, KIF2A, KIF5C, KMT2E, KRAS, LMBRD2, LMNB1, MACF1, MAF, MAP2K1, MAP2K2, MAST1, MAST3, MAST4, MBD5, MINPP1, MTHFR, MTOR, NARS1, NBEA, NCDN, NDE1, NEDD4L, NEUROD2, NR4A2, NSD1, NSF, OTX2, PAFAH1B1, PEX6, PIGT,*	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Epilepsy_v2025_Tier1, Syndrom_Epilepsy_v2025_Tier2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
PIK3R2, PNPT1, POLG2, PPP2CA, PPP3CA, PRODH, PRPF8, PSAP, PTEN, PUM1, PURA, RAB11B, RAC3, RALA, RANBP2, RELN, RNF13, RNF2, RNU4-2, RORA, RORB, RRM2B, RYR2, SAMHD1, SATB1, SATB2, SCO1, SCO2, SDHA, SERPINI1, SETBP1, SETD5, SGSH, SIK1, SIX3, SLC12A5, SLC2A1, SLC32A1, SLC6A1, SMARCA2, SMARCC2, SNAP25, SPTAN1, SPTBN1, STAG1, STARD7, SYNGAP1, TANC2, TBC1D24, TBL1XR1, TCF4,, TMEM106B, TMEM63B, TNPO2, TRAF7, TREX1, TRPM3, TRRAP, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, U2AF2, UBAP2L, UBE3A, UFSP2, USP7, ZBTB18, ZEB2, ZMIZ1, ZMYM2, ALG13, ARHGEF9, ATP6AP2, ATP7A, ATRX, CASK, CLCN4, CUL4B, DCX, DDX3X, EIF2S3, FAM50A, FLNA, GLRA2, HCCS, HCFC1, HNRNPH2, 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, BSCL2, CAPRIN1, CSNK1G1, DROSHA, EMX2, GNAQ, HEATR5B, JAKMIP1, KCND2, KCTD3, KLHL20, PARP6, RAB11A, SCAF4, SCAMP5, SNX27, SPR, SYNCRIP, TNK2, TXNRD1, WNK3, AARS1, AASS, ABCA2, ADAM22, ADAR, ADAT3, ADGRG1, ADPRS, ADSL, AIMP1, ALDH5A1, ALG1, ALG11, ALG14, ALG3, ALG6, ALG9, ALKBH8, AMPD2, AP1G1, APC2, ARFGEF2, ARG1, ASAH1, ASNS, ATP5O, ATP6V0A2, BCKDHA, BCKDHB, BCS1L, BLOC1S1, BLTP1, BRAT1, BTD, C12orf57, C2orf69, CACNA2D1, CACNA2D2, CARS2, CAMSAP1, 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, ESAM, 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, GRM7, GSS, GTPBP2, GTPBP3, GUF1, HACE1, HAX1, HECTD4,				

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
HERC2, HEXA, HEXB, HID1, HLCS, HMGCL, HOXA1, HPDL, HSD17B4, HTRA2, IER3IP1, KARS1, KATNB1, KCNJ10, KIFBP, KPTN, LARGE1, LARS1, LETM1, LIAS, LIPT1, LIPT2, LMAN2L, LNPK, LSS, LYST, MADD, MAGI2, MANBA, MBOAT7, MED11, 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, OTUD7A, OXR1, P4HTM, PAH, PCCA, PCCB, PCDH12, PCDHGC4, PCLO, PCYT2, PDHX, PDSS2, PET100, PEX1, PEX10, PEX12, PEX13, PEX19, PEX2, PEX3, PEX5, PEX7, PGM2L1, PHGDH, PI4K2A, PIDD1, PIGC, PIGG, PIGK, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGU, PIGW, PLA2G6, PLAA, PLCB1, PMM2, PMPCB, PNKP, POLG, POMGNT1, POMT1, POMT2, PPFIBP1, PPIL1, PPT1, PRICKLE1, PRMT7, PSAT1, PSPH, PTCD3, PTF1A, PTPN23, PTS, QARS1, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RALGAPA1, RARS1, RARS2, RFT1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPIA, RTN4IP1, RTTN, RUSC2, SARS1, SEPSECS, SHQ1, SLC1A4, SLC13A3, SLC25A1, SLC31A1, SLC35A1, SLC35A3, SLC39A8, SLC4A10, SLC45A1, SLC5A6, SNIP1, SNORD118, SPATA5, SPATA5L1, SPTBN4, ST3GAL3, ST3GAL5, STAMBP, STRADA, SUCLA2, SUCLG1, SUOX, SURF1, TAF8, TANGO2, TBC1D20, TBCD, TDP2, TELO2, TIMM50, TMEM222, TMEM70, TMX2, TPP1, TRAPPC12, TRAPPC6B, TRIP13, TRIT1, TRPM6, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TUBGCP2, UBR7, UFC1, UFM1, UNC80, USP18, VARS1, VLDLR, VPS11, VPS50, WARS2, WDR45B, WDR62, WDR73, YIF1B, YIPF5, ZNF335); SNV, CNV				
Autismus (*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,*	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Autism_v2024_Tier1 und Syndrom_Autism_v2024_Tier2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
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, 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, 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); SNV, CNV				

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Marfan-Syndrom (*FBN1, TGFBR1, TGFBR2, IPO8, COL1A2, FKBP14*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_marfan_v2023, Pipeline V.2	30.05.2024
Loeys-Dietz-Syndrom (*COL3A1, IPO8, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Loeys-Dietz_v2024, Pipeline V.2	30.05.2024
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, COPB2, MBTPS2, NUDT6, SGMS2, SUCO*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Other_Osteogenesis_imperfecta_Tier1_v2023, Pipeline V.2	30.05.2024
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, 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, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Joubert_v2023, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Retinitis Pigmentosa (*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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensorineural_Retinitis_pigmentosa_v2024_Tier1 und Sensorineural_Retinitis_pigmentosa_v2024_Tier2 ,Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensorineural_Stargardt_v2024_Tier1+2, Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensorineural_Zapfen-Stäbchen-Dystrophie_v2024_only1Tier, Pipeline V.2	30.05.2024
Retinale Erkrankungen (*ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, AFG3L2, AGBL5, AHI1, 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,*	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Retinale Erkrankungen_v2024_only1Tier, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
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, MTTP, 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, 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); SNV, CNV				
Okulärer und okulokutaner Albinismus (*GPR143, HPS1, HPS3, HPS4, HPS5, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Albinism_v.2024_only1Tier, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Senior-Loken-Syndrom (*CEP290, IQCB1, NPHP1, NPHP3, NPHP4, POC1B, SDCCAG8, TRAF3IP1, WDR19*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Optikusatrophie_v.2025_only1Tier_Pipeline V.2	30.05.2024
Optikusatrophie (*ACO2, AFG3L2, ALPK1, DNM1L, MFN2, NR2F1, OPA1, OPA3, SPG7, SSBP1, WFS1, MT-ND1, MT-ND4, MT-ND6, CISD2, DNAJC30, FDXR, MECR, NBAS, NDUFA12, PDXK, RTN4IP1, SLC25A46, TMEM126, ATAD3A, ATP1A3, C19orf12, HK1, LHX2, LRRC8C, MIEF1, POLG, UCHL1, HSD17B10, TIMM8A , MT-ATP6, AP3B2, ATG7, BLOC1S1, BORCS8, BTD, EPRS1, HIKESHI, ISCA2, LETM1, MAG, MCAT, MFF, MTRFR, PDSS1, SLC44A1, SC52A2, SNF8, TFG, YME1L1); SNV, CNV*	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Optikusatrophie_v.2025_Tier1, Optikusatrophie_v.2025_Tier2, Pipeline V.2	17.10.2025
Kardiomyopathien (*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, 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,*	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_Kardiomyopathien-breit_v2024_Tier1 und Kardio_Kardiomyopathien-breit_v2024_Tier2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
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				
HCM (*MYL3, MYH7, PLN, FLNC, ACTC1, ACTN2, CSRP3, LAMP2, MYBPC3, MYL2, TNNC1, TNNI3, TNNT2, TPM1, TTR, GLA, ALPK3, CACNA1C, FHL1, FHOD3, PRKAG2, TRIM63, FXN, 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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_HCM_v2024_Tier1 und Kardio_HCM_v2024_Tier2, Pipeline V.2	30.05.2024
DCM (*ACTC1, ACTN2, BAG3, CDH2, DSC2, DSG2, DSP, JUP, LMNA, MYBPC3, MYH6, MYH7, NEXN, NKX2-5, PLN, PRDM16, RBM20, SCN5A, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, DMD, LAMP2, DOLK, EPG5, ABCC9, ANK2, ANKRD1, CRYAB, CSPR3, CTF1, DTNA, EYA4, FLNC, GATA6, IDH2, ILK, JPH2, LAMA4, LBD3, MYL2, MYLK3, MYPN, NEBL, NRAP, PKP2, PSEN1, RAF1, RRAGC, RRAGD, RYR2, SCN1B, SGCD, TAB2, TBX5, TBX20, TCAP, TTR, FKRP, FKTN, FLII, GATAD1, MYZAP, RHBDF1, RPL3L, SLC6A6, SPEG*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_DCM_v2025_Tier1 und Kardio_DCM_v2025_Tier2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Arrhythmien (*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, 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, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_Arrhythmia_v2024_Tier1 und Kardio_Arrhythmia_v2024_Tier2, Pipeline V.2	30.05.2024
Angeborene Herzfehler nicht syndromal (*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, commercial pipeline	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	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Angeborene Herzfehler breit (*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, RBFOX2, RRAS2, SALL1, SETBP1, SLC2A10, SOS2, TRAF7, ACVR2B, CITED2, CRELD1, GATA5, MEIS2, MYH6, NKX2-6, SMAD6, TLL1, ZFPM2, ZIC3, ABCC9, ADAMTS10, ADAMTS19, ADNP, AFF4, ALX3, ARHGAP31, ARID1B, ASXL2, ATRX, B3GALT6, 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, 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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	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	30.05.2024
TAAD (*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, TGFBR1, TGFBR2; CBS, FLCN, FOXE3, HCN4, MAT2A, MFAP5, SKI*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_TAAD_v2024 Tier 1 und Kardio_TAAD_v2024 Tier 2, Pipeline V.2	30.05.2024
PAH (*ACVRL1, ATP13A3, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNK3, KDR, SMAD9, SOX17, TBX4, ABCC8, AQP1, BMP10, BMPR1A, BMPR1B, FBLN2, GGCX, KLF2, KLK1, NFU1, NOTCH3, PDGFD, SARS2, SMAD1, SMAD4, TET2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_PAH_v2024_Tier1 und Kardio_PAH_v2024_Tier2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Long_QT (*CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN, ANK2, CAV3, KCNE2, KCNJ5, SCN4B, SNTA1, TECRL*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_LongQT_v2025_Tier1 und Kardio_LongQT_v2025_Tier2, Pipeline V.2	30.05.2024
Short QT (*CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3, SCN5A*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_ShortQT_v2025_only1Tier, Pipeline V.2	17.10.2025
ARVC (*DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, TMEM43, ANK2, CAV3, CDH2, CTNNA3, MYH7, MYL3, PLN, SCN5A, TGFB3, TJP1, TTN*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_ARVC_v2025_Tier1 und Kardio_ARVC_v2025_Tier2, Pipeline V.2	30.05.2024
Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) (*CALM1, CALM2, CALM3, RYR2, CASQ2, TECRL, TRDN*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, CPVT v2025 nur1Tier, Pipeline V.2	17.10.2025
Brugada (*CACNA1C, CACNB2, SCN5A, GPD1L, HCN4, KCND3, KCNE3, KCNH2, SCN1B, SCN3B, TRPM4*) ; SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Kardio_Brugada_v2025_Tier1 und Kardio_Brugada_v2025_Tier2, Pipeline V.2	17.10.2025
Meckel-Gruber-Syndrom (*B9D1, B9D2, CC2D2A, CEP290, CSPP1, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TCTN3, TMEM107, TMEM216, TMEM231, TMEM67, TXNDC15, WDPCP*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Meckel-Gruber_v2023, Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Polyzystische Nierenerkrankung_v2025, Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Syndrom_Primäre ziliäre Dyskinesie, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Sensorineurale Schwerhörigkeit (*GJB2, GJB6*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Sensineural_Hearingloss_GJB2/6, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Sensorineurale Schwerhörigkeit (*ACTG1, ATP2B2, ATP6V1B2, CCDC50, CEACAM16, COCH, COL11A1, COL11A2, COL2A1, CRYM, DFNA5, DIAPH1, DMXL2, DSPP, EDN3, EDNRB, ESPN, EYA4, GATA3, GJB2, GREB1L, GRHL2, HOXA2, KCNQ4, LMX1A, MITF, MYH14, MYH9, MYO6, MYO7A, OSBPL2, P2RX2, PLS1, POU4F3, PTPRQ, SLC17A8, SOX10, TBC1D24, TECTA, TMC1, USP48, WFS1, MT-RNR1, AIFM1, COL4A5, POU3F4, PRPS1, SMPX, TIMM8A , ADGRV1, BSND, CABP2, CDC14A, CDH23, CIB2, CISD2, CLDN14, CLDN9, CLPP, CLRN1, COL9A1, DAP3, DFNB59, EPS8, EPS8L2, ESRRB, FGF3, GGPS1, GIPC3, GPR156, GPSM2, GRXCR1, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNJ16, KCNQ1, KIAA1024L, LARS2, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MPZL2, MSRB3, MYO15A, MYO3A, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PKHD1L1, RDX, RNF220, S1PR2, SERPINB6, SLC26A4, SLC26A5, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SPATA5, SPATA5L1, STRC, SYNE4, TMIE, TMPRSS3, TPRN, TRIOBP, USH1C, USH1G, USH2A, WHRN, ABCC1, ACOX1, ATP11A, BMP4, CHD7, COG4, DIABLO, DIAPH3, DNMT1, EDNRA, EFTUD2, ELMOD3, EYA1, FGF10, FGFR2, FGFR3, FOXI3, GDF6, GJB3, GJB6, GNAI3, GSDME, HOMER2, KDM3B, KIT, KITLG, KMT2D, MCM2, MIR96, MN1, MORC2, NLRP12, OPA1, OTX2, PAX2, PAX3, PBX1, PLCB4, PMP22, POLR1A, POLR1D, REST, RIPOR2, RPS28, SALL1, SALL4, SCD5, SF3B4, SIX1, SIX5, SLC12A2, SNAI2, SOX2, SPATC1L, STXBP3, TCOF1, TFAP2A, THOC1, TMTC2, TNC, TOP2B, TUBB4B, TWNK, MT-TS1, COL4A6, KDM6A, OFD1, ABHD12, AFG2A, ALMS1, AP1S1, ATP6V1B1, BCS1L, CDC6, CDT1, CEP250, CEP78, CLIC5, CLRN2, COL9A2, COL9A3, CRLS1, DHODH, DNAJC3, EIF4A3, ESRP1, EXOSC2, FDXR, FOXF2, FRAS1, FREM2, GRIP1, GRXCR2, GSC, HAAO, HARS2, HGF, HMX1, HSPA9, IARS2, KARS1, LETM1, MASP1, MET, NARS2, OGDHL, ORC1, ORC4, ORC6, OXR1, PDSS1, PHYH, PJVK, PLXNB2, PNPT1, POLR1C, PPIP5K2, RFC4, ROR1, SERAC1, SGPL1, SPNS2, SPTBN4, STX4, WBP2, YARS*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Hörstörung_v2025_Tier1, Hörstörung_v2025_Tier2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Familiärer Brustkrebs (*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53, ATRIP, CDKN1B, CDKN2A, CTNNA1, EPCAM, MLH1, MSH2, MSH6, NBN (nur rs587776650), NF1, PMS2, POLD1, POLE, PTCH1, MUTYH, FANCB, BLM, ERCC3, ERCC4, ERCC5, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, NTHL1, POLH, SLX4, UBE2T, WRN*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Brustkrebs_v2025_Tier1 und Tumordisp_Brustkrebs_v2025_Tier2, Pipeline V.2	17.10.2025
Familiärer Eierstockkrebs (*BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, STK11, TP53, APC, ATM, CDKN2A, CHEK2, DICER1, FH, LLGL2, POLD1, POLE, PTCH1, PTEN, SMARCA4, SMARCB1, SUFU, TRIM37, WT1, MUTYH, FANCB, BLM, ERCC3, ERCC4, ERCC5, NTHL1, POLH, SLX4, TOP3A, TRIM37, UBE2T, WRN, XPA, XPC, XRCC2);* SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Eierstockkrebs_v2025_Tier1 und Tumordisp_Eierstockkrebs_v2025_Tier2, Pipeline V.2	17.10.2025
Familiärer Brust- und Eierstockkrebs (*BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53, APC, ATRIP, CDKN1B, CDKN2A, CTNNA1, DICER1, EPCAM, LLGL2, NBN, NF1, POLD1, POLE, PTCH1, SMARCA4, SMARCB1, SUFU, WT1, MUTYH, FANCB, BLM, ERCC3, ERCC4, ERCC5, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, NTHL1, POLH, SLX4, TRIM37, UBE2T, WRN);* SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_HBOC_v2025_Tier1 und Tumordisp_HBOC_v2025_Tier2, Pipeline V.2	30.05.2024
Familiärer Brust- und Eierstockkrebs (Therapierelevanz) (*BRCA1, BRCA2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_BRCA1/2_only, Pipeline V.2	30.05.2024
Li-Fraumeni-Syndrom (*CHEK2, POT1, TP53, APC, BRCA1, BRCA2, CDH1, GPR161, HRAS, KRAS, MAX, MEN1, MLH1, MSH2, NBN, NF1, NF2, NRAS, PALB2, PMS2, POLD1, POLE, PRKAR1A, PTCH1, PTEN, PTPN11, RAD51C, RAD51D, RAF1, RB1, REST, RET, RIT1, RNF43, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SMAD4, SMARCA4, SMARCB1, SOS1, STK11, SUFU, TERC, TERT, TMEM127, TSC1, TSC2, VHL, WT1, GPC3, GNAS, MUTYH, NTHL1,RECQL4, SLX4, TRIM37, XRCC2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_LiFraumeni_v2025 Tier 1 und Tumordisp_LiFraumeni_v2025 Tier 1, Pipeline V.2	30.05.2024
Lynch-Syndrom (*MLH1, PMS2, MSH6, MSH2, EPCAM*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Lynch-syndrom_v2025, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Polyposis (*APC, AXIN2, BMPR1A, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, GREM1, MUTYH, NTHL1, EPCAM, MLH1, MSH2, MSH6, PMS2, FANCB, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, FANCG, FANCL, MBD4, MSH3, SLX4, UBE2T, XRCC2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	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	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Peutz-Jeghers_v2023, Pipeline V.2	30.05.2024
Neurofibromatose / Schwannomatose (*LZTR1, NF1, NF2, PRKAR1A, SMARCB1, SPRED1, BRAF, BRCA2, BRIP1, EPCAM, GNAS, HOXB13, KITLG, MAP2K1, PDGFRB, PTPN11, RAF1, RIT1, SOS1, FANCB, POLA1, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, SLX4*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Neurofibromatosis_v.2025_Tier1 und Tumordisp_Neurofibromatosis_v.2025_Tier2, Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Cowden_v2023, Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Gorlin-Goltz_v2023, Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Birt-Hogg-Dubé_v2023, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_VHL _v2023, Pipeline V.2	30.05.2024
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Multiple Exostosen_v2023, Pipeline V.2	30.05.2024
Multiple endokrine Neoplasie (*CDKN1B, MEN1, RET, CDC73, FH, MAX, NF1, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, UBE2T, TP53, AIP, CASR, DLST, DNMT3A, MDH2, SLC25A11, VHL*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_MEN_v.2024_Tier1 und Tumordisp_MEN_v.2024_Tier2, Pipeline V.2	30.05.2024
Hereditäres Phäochromozytom-Paragangliom-Syndrom (*DLST, FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL, SLC25A11, EGLN1, EPAS1, MERTK, MET, MDH2, PRKAR1A, SUCLG2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_PPGL_v2025_Tier1 und Tumordisp_PPGL_v2025_Tier2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Solide kindliche Tumore (*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, 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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Kindliche-Tumore_v2024_Tier1 und Tumordisp_Kindliche-Tumore_v2024_Tier2, Pipeline V.2	30.05.2024
Pankreaskarzinom (*ATM, BRCA1, BRCA2, CDKN2A, MLH1, MSH2, MSH6, PMS2, PALB2, STK11, TP53, APC, BMPR1A, EPCAM, PRSS1, PTEN, SMAD4, SPINK1, VHL*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Pankreaskarzinom_v.2025 Tier1 und Tumordisp_Pankreaskarzinom_v.2025 Tier2, Pipeline V.2	30.05.2024
Blasen- und Harnleiterkrebs (*MLH1, MSH2, MSH6, PMS2, BAP1, EPCAM, FH, MUTYH, NF1, SMARCA4, SMARCB1, STK11, TP53, NTHL1*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	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	30.05.2024
Endometriumkarzinom (*EPCAM, FH, MLH1, MSH2, MSH6, PMS2, POLD1, POLE, PTEN, SMAD4, MUTYH, NTHL1*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_EndometriumCA_v2025_only1Tier, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
GIST (*KIT, PDGFRA, SDHA, SDHB, SDHC, SDHD, APC, BMPR1A, CDH1, CTNNA1, MLH1, MSH2, MSH6, MBD4, MUTYH , NTHL1*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_GIST_v2025_Tier1, Tumordisp_GIST_v2025_Tier2, Pipeline V.2	30.05.2024
Magenkrebs (*APC, BMPR1A, CDH1, CHEK2, CTNNA1, EPCAM, MLH1, MSH2, MSH6, PDGFRA, PMS2, POLD1, POLE, PTPN11, SMAD4, STK11, TP53*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Magenkrebs_v2025_only1Tier, Pipeline V.2	30.05.2024
Medulloblastom (*APC, BRCA2, DICER1, ELP1, GPR161, PALB2, PTCH1, RET, SEC23B, SUFU, TP53, GPC3, NBN*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Medulloblastom_v.2024_only1Tier, Pipeline V.2	30.05.2024
Schilddrüsenkarzinom (*APC, CDKN1B, CHEK2, DICER1, FLCN, PRKAR1A, PTEN, RET, TP53, CDC73, FOXE1, GNAS, MSH2, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, TSC1, TSC2, MUTYH, SLC5A5, WRN*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Schilddrüsen CA_v2025_Tier1 und Tumordisp_Schilddrüsen CA_v2025_Tier2, Pipeline V.2	30.05.2024
Tuberöse Sklerose (*TSC1, TSC2, 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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Tuberöse_Sklerose_v.2024_Tier1 und Tumordisp_Tuberöse_Sklerose_v.2024_Tier2, Pipeline V.2	30.05.2024
Nierenkarzinom (*BAP1, CDC73, CDKN2B, CHEK2, DICER1, FH, MET, PTEN, REST, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, TSC1, TSC2, VHL, WT1, CDKN1B, CTR9, EPCAM, FLCN, MLH1, MSH2, MSH6, PMS2, STK11, DIS3L2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Nierenkarzinom_v.2024, Pipeline V.2	30.05.2024
Sarkoma (*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, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Sarcoma_v.2024_Tier1 und Tumordisp_Sarcoma_v.2024_Tier2, Pipeline V.2	30.05.2024
Melanom (*ACD, BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN, RB1, TERT, TERF2IP, CYLD, FLCN, LEMD3, MC1R, PRDM10, PTCH1, TP53, GNAS, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, RECQL4, WRN, XPA, XPC*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Melanome_v.2025, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Prostatakarzinom (*ATM, BRCA1, BRCA2, CHEK2, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, CDKN1B, EPCAM, GNAS, RAD51D, TP53); SNV, CNV*	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_ProstataCA_v2025 Tier1, Tumordisp_ProstataCA_v2025 Tier2, Pipeline V.2	17.10.2025
Hirntumorpanel (*APC, ATM, BRCA2, CDNK2A, 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, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_Hirntumor_v2025_only1Tier, Pipeline V.2	17.10.2025
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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Tumordisp_alleGene_Tier1+2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Primäre Ovarialinsuffiziens 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, EIF2B4, GDF9, NOG, NUP107, POU5F1, RCBTB1, SOHLH2, BMPR1B, BUB1B, DACH2, EIF4ENIF1, FIGLA, LMNA, MSH5, NANOS3, PGRMC1, POF1B, POLR2C, POLR3H, PSMC3IP, SGO2, SYCP2L, KHDRBS1, 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, 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*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Other_Primary_ovarial_insufficiency_v.2024_Tier1, Pipeline V.2	30.05.2024
Erbliche Blutungserkrankungen (*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), LAT, ORAI1, PTPRJ, SLC35A1, TPM4, VPS33B, VWF, WAS*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Other_Inherited_bleeding_disorders_v.2024_Tier 1 und Other_Inherited_bleeding_disorders_v.2024_Tier 2, Pipeline V.2	30.05.2024

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	aufgenommen am
Leukodystrophie (*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, NASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RPS6KA3, SAMHD1, SNORD118, SPG11, SPG21, TREM2, TREX1, TTR, TUBB4A, TYMP, TYROBP, ZFYVE26, MLC1, CST3, CTC1, EARS2, GCDH, KIF5A, MAL, NPC1, PSEN2, TPP2*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Other_Leukodystrophy_adultonset Tier1_v.2024 und Other_Leukodystrophy_adultonset Tier2_v.2024, Pipeline V.2	30.05.2024
MODY/neonataler Diabetes (*ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCN11, NEUROD1, PDX1, RFX6, CNOT1, EIF2B1, GATA4, GATA6, INSR, STAT3, WFS1, EIF2S3, FOXP3, KLF11, BSCL2, EIF2AK3, FICD, GLIS3, IER3IP1, IL2RA, LBRA, MNX1, NEUROG3, NKX2-2, ONECUT1, PAX1, PDIA6, PTF1A, SLC19A2, SCL2A2, YIPF5, ZFP57, ZNF808*); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina PCR-Free Library Prep V.3, Other_MODY_Tier1_v.2025 und Other_MODY_Tier2_v.2025, Pipeline V.2	17.10.2025
Segregationsanalysen oder Untersuchung auf Anlageträgerschaften (z.B. *ABCC8, BRCA1, BRCA2, BRPF1, DARS2, DICER1, FIG4, MYBPC3, NCDN, NONO, PTPN11, TNNT2*)	EDTA-Blut, DNA; DNA	PCR und Sanger-Sequenzierung	GEN-AA-Sanger-Sequenzierung V.1	15.11.2023
Therapieindikation Olaparib (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) (*BRCA1, BRCA2*); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, commercial pipeline	GEN-AA-Illumina Exome 2.5 V.1, Tumordisp_BRCA1/2_only, Pipeline V.2	15.11.2023
Sensorineurale Schwerhörigkeit (GJB2)	EDTA-Blut, DNA; DNA	PCR und Sanger-Sequenzierung	GEN-AA-Sanger-Sequenzierung V.3	15.11.2023