

## Deutsche Akkreditierungsstelle

### Anlage zur Akkreditierungsurkunde D-ML-22580-01-00 nach DIN EN ISO 15189:2024

**Gültig ab:** 12.06.2025

Ausstellungsdatum: 12.06.2025

Inhaber der Akkreditierungsurkunde:

**Medizinisches Versorgungszentrum am Klinikum Oldenburg GmbH  
Rahel-Straus-Straße 10, 26133 Oldenburg**

mit dem Standort

**Medizinisches Versorgungszentrum am Klinikum Oldenburg GmbH  
Medizinische Genetik  
Rahel-Straus-Straße 10, 26133 Oldenburg**

Das Medizinische Laboratorium erfüllt die Anforderungen gemäß DIN EN ISO 15189:2024, um die in dieser Anlage aufgeführten Konformitätsbewertungstätigkeiten durchzuführen. Das Medizinische Laboratorium erfüllt gegebenenfalls zusätzliche gesetzliche und normative Anforderungen, einschließlich solcher in relevanten sektoralen Programmen, sofern diese nachfolgend ausdrücklich bestätigt werden.

Die Anforderungen an das Managementsystem in der DIN EN ISO 15189 sind in einer für medizinische Laboratorien relevanten Sprache verfasst und stehen insgesamt in Übereinstimmung mit den Prinzipien der DIN EN ISO 9001.

*Diese Urkundenanlage gilt nur zusammen mit der schriftlich erteilten Urkunde und gibt den Stand zum Zeitpunkt des Ausstellungsdatums wieder. Der jeweils aktuelle Stand der gültigen und überwachten Akkreditierung ist der Datenbank akkreditierter Stellen der Deutschen Akkreditierungsstelle zu entnehmen ([www.dakks.de](http://www.dakks.de))*

**Untersuchungen im Bereich:**

Medizinische Laboratoriumsdiagnostik

**Untersuchungsgebiet:**

Humangenetik (Molekulare Humangenetik)

**Flexibler Akkreditierungsbereich:**

Dem Medizinischen Laboratorium ist innerhalb der gekennzeichneten Untersuchungsbereiche, ohne dass es einer vorherigen Information und Zustimmung der DAkKS bedarf,

[Flex C] die Modifizierung sowie Weiter- und Neuentwicklung von Untersuchungsverfahren gestattet.

Die aufgeführten Untersuchungsverfahren sind beispielhaft. Das Medizinische Laboratorium verfügt über eine aktuelle Liste aller Untersuchungsverfahren im flexiblen Akkreditierungsbereich. Die Liste ist öffentlich verfügbar auf der Webpräsenz des Medizinischen Laboratoriums.

## Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

### Untersuchungsart:

### Molekularbiologische Untersuchungen [Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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, in-house pipeline
Fluoropyrimidin Toxizität (DPD-Gen: dbSNP rs3918290, rs55886062, rs67376798, rs56038477)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler
Morbus Bechterew (HLA-B27 Subtypen B*2702 und B*2705)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler
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
Thrombophilie (F5-Gen: dbSNP rs6025, F2-Gen: rs1799963)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler
Hämochromatose (HFE-Gen: rs1800562, rs1799945)	EDTA-Blut, DNA; EDTA-Blut, DNA	Loop-mediated isothermal amplification, Real-time PCR Cycler
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
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
Noonan Syndrom/RASopathie ( <i>BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Aicardi-Goutières-Syndrom ( <i>ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Bardet-Biedl-Syndrom ( <i>ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TTC8, WDPCP</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
CHARGE-Syndrom ( <i>CHD7</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Cornelia-de-Lange-Syndrom ( <i>HDAC8, NIPBL, RAD21, SMC3</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Ehlers-Danlos Syndrom ( <i>ADAMTS2, ADAMTSL2, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, DSE, FKBP14, PLOD1, SLC39A13, AEBP1, COL5A1, COL5A2, TNXB, COL3A1</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Coffin-Siris-Syndrom ( <i>ARID1A, ARID1B, BICRA, HDAC8, NIPBL, PHF6, PIGV, RAD21, SMARCA2, SMARCA4, SMC3, TBC1D24</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Kabuki-Syndrom ( <i>CHD7, EYA1, FLNB, IRF6, KDM6A, KMT2D, SIX1, SIX5</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Rubenstein-Taybi-Syndrom ( <i>CREBBP, EP300, FGFR1, FGFR2, GLI3, HOXD13, SRCAP, TWIST1</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Smith-Lemli-Opitz-Syndrom ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>Epilepsie Tier 1 (AFF3, AP2M1, ATP6V1A, CACNA1A, CACNA1E, CACNA1C, CACNA1D, CACNA1G, CACNA1I, CDK19, CELF2, CHD2, CHRNA4, CHRNB2, CNNM2, CSNK2B, CYFIP2, DEPDC5, DHDDS, DNM1, EEF1A2, FBXO28, FGF12, GABBR2, GABRA1, GABRA2, GABRA5, GABRB2, GABRB3, GABRG2, GNAO1, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCN1, HCN2, HECW2, HNRNPU, KCNB1, KCNC1, KCNC2, KCNQ2, KCNQ3, KCNT1, KCNT2, LGI1, MEF2C, NACCC1, NPRL2, NPRL3</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>
<p>Epilepsie Tier 1 (NTRK2, NUS1, PACS1, PACS2, PAK1, PHACTR1, PRRT2, RHOBTB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SETD1A, SETD1B, SIK1, SLC1A2, STX1B, STXBP1, TRAPPC4, TRIM8, VAMP2, WASF1, WDR37, YWHAG, ARX, CDKL5, CNKSR2, FGF13, OPHN1, PCDH19, SYN1, ABAT, ADARB1, ALDH7A1, AP3B2, ARV1, CACNA1B, CAD, CERS1, CNPY3, CNTNAP2, CSTB, CTNNA2, D2HGDH, DENND5A, DOCK7, EPM2A, FOLR1, FRRS1L, GOSR2, GOT2, GPAA1, ITPA, KCTD7, MDH2, MTHFS, NAPB, NHLRC1, NRROS, PARS2, PIGB, PLPBP, SCARB2, SLC13A5, SLC25A12, SLC38A3, SYNJ1, SZT2, TBC1D2B, TIAM1, TRAK1, UBA5, UGDH, UGP2, ZNF142); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Epilepsie Tier 2 (ACOX1, ACTL6B, AIMP2, AKT3, ALG8, ALPL, ANKRD11, ARF1, ARFGEF1, ARID1B, ASH1L, ASPA, ASXL3, ATN1, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5F1A, ATP6V0A1, ATP6VOC, BAP1, BOLA3, TET3, BRAF, BSCL2, CACNA1H, CCDC88C, CDC42BPB, CEP85L, CHD4, CHD5, CHRNA2, CIC, CLCN3, CLPB, CLTC, COG4, COG8, COL18A1, COL4A1, COL4A2, CPA6, CREBBP, CTU2, CUL3, CUX2, DEAF1, DHX16, DLL1, DNAJC5, DNM1L, DYNC1H1, DYRK1A, EFHC1, EFTUD2, EHMT1, EIF2AK2, EXT2, FAR1, FBXO11, FGFR3, FH, FOXG1, FRMD5, GABRB1, GABRD, GBA, GCH1, GFAP, GLI3, GLRA1, GLS, GLUD1, GNB1, GPHN, GRIA2, GRN)	EDTA-Blut, DNA; DANN	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Epilepsie Tier 2 (H3-3A, H3-3B, HEPACAM, HNRNPR, HRAS, HSPD1, IFIH1, IRF2BPL, KAT5, KAT8, KCNA1, KCNA2, KCNH1, KCNH5, KCNJ11, KCNK4, KCNMA1, KCNQ5, KIF1A, KIF2A, KIF5C, KMT2E, KRAS, LMBRD2, LMNB1, MACF1, MAF, MAP2K1, MAP2K2, MAST1, MBDS, MINPP1, MTHFR, MTOR, NARS1, NBEA, NCDN, NDE1, NEDD4L, NEUROD2, NR4A2, NSD1, NSF, OTX2, PAFAH1B1, PEX6, PIGT, PIK3R2, PNPT1, POLG2, PPP2CA, PPP3CA, PRODH, PRPF8, PSAP, PTEN, PUM1, PURA, RAB11B, RALA, RANBP2, RELN, RNF13, RNF2, RORA, RORB, RRM2B, RYR2, SAMHD1, SATB1, SATB2, SCO1, SCO2, SDHA, SERPINI1, SETBP1, SETD5, SGSH, SIX3, SLC12A5, SLC2A1, SLC6A1, SMARCA2, SMARCC2, SNAP25, SPTAN1, SPTBN1, STAG1, STARD7, SYNGAP1)	EDTA-Blut, DNA; DANN	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Epilepsie Tier 2 (TANC2, TBC1D24, TBL1XR1, TCF4, TMEM106B, TNPO2, TRAF7, TREX1, TRPM3, TRRAP, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, UBE3A, UFSP2, USP7, ZBTB18, ZEB2, ZMIZ1, ZMYM2, ALG13, ARHGEF9, ATP6AP2, ATP7A, ATRX, CASK, CLCN4, CUL4B, DCX, DDX3X, EIF2S3, FAM50A, FLNA, GLRA2, HCCS, HCFC1, 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, CAPRIN1, CSNK1G1, DROSHA, EMX2, GNAQ, HEATR5B, JAKMIP1, KCND2, KCTD3, KLHL20, PARP6, RAB11A, SCAF4, SCAMP5, SNX27, SPR, SYNCRIP, TNK2, TXNRD1, WNK3)	EDTA-Blut, DNA; DANN	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>Epilepsie Tier 2 (AARS1, ABCA2, ADAM22, ADAR, ADAT3, ADGRG1, ADPRS, ADSL, AIMP1, ALDH5A1, ALG1, ALG11, ALG14, ALG3, ALG6, ALG9, ALKBH8, AMPD2, AP1G1, APC2, ARFGEF2, ARG1, ASAH1, ASNS, ATP6V0A2, BCKDHA, BCKDHB, BCS1L, BLOC1S1, BLTP1, BRAT1, BTB, C12orf57, CACNA2D1, CACNA2D2, CARS2, CC2D2A, CCDC88A, CHKA, CLN3, CLN5, CLN6, CLN8, CNTN2, COG6, COG7, COLGALT1, COQ2, COQ4, COQ6, COQ9, COX10, COX15, CPLX1, CPSF3, CRPPA, CTSD, CTSF, CYP27A1, DBT, DDC, DEGS1, DHCR24, DHCR7, DHPS, DHX30, DIAPH1, DMXL2, DNAJC6, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPYD, DTYMK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF3F, EMC10, EML1, EPG5, ETHE1, EXOC7, EXOSC3, FARS2, FASTKD2, FBXL4, FCSK, FDFT1, FKRP, FKTN, FOXRED1, FUCA1, FUT8, GAD1, GALC, GALNT2, GAMT, GCSH, GFM1, GLB1, GLDC, GLUL, GLYCTK, GM2A, GNB5, GSS, GTPBP2, GTPBP3, GUF1)</p>	<p>EDTA-Blut, DNA; DANN</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Epilepsie Tier 2 (HACE1, HAX1, HERC2, HEXA, HEXB, HID1, HLCS, HMGCL, HOXA1, HPDL, HSD17B4, HTRA2, IER3IP1, KARS1, KATNB1, KCNJ10, KIFBP, KPTN, LARGE1, LARS1, LIAS, LIPT1, LIPT2, LMAN2L, LNPk, LSS, LYST, MADD, MAGI2, MANBA, MBOAT7, MED17, MED27, MFF, MFSD8, MLC1, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MTR, NAGA, NARS2, NDUFA10, NDUFA2, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NGLY1, NRXN1, NSRP1, NUBPL, NUP214, OCLN, OGDHL, OTUD6B, OXR1, P4HTM, PAH, PCCA, PCCB, PCDH12, PCDHGC4, PCLO, PCYT2, PDHX, PDSS2, PET100, PEX1, PEX10, PEX12, PEX13, PEX19, PEX2, PEX3)	EDTA-Blut, DNA; DANN	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>Epilepsie Tier 2 (PEX5, PEX7, PGM2L1, PHGDH, PID1, PIGC, PIGG, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGU, PIGW, PLA2G6, PLAA, PLCB1, PMM2, PMPCB, PNKP, POLG, POMGNT1, POMT1, POMT2, PPIL1, PPT1, PRICKLE1, PRMT7, PSAT1, PSPH, PTF1A, PTPN23, PTS, QARS1, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RALGAPA1, RARS1, RARS2, RFT1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPIA, RTN4IP1, RTTN, RUSC2, SARS1, SEPSECS, SLC1A4, SLC25A1, SLC31A1, SLC35A1, SLC35A3, SLC39A8, SLC45A1, SLC5A6, SNIP1, SNORD118, SPATA5, SPATA5L1, SPTBN4, ST3GAL3, ST3GAL5, STAMBIP, STRADA, SUCLA2, SUCLG1, SUOX, SURF1, TAF8, TANGO2, TBC1D20, TBCD, TDP2, TELO2, TIMM50, TMEM222, TMEM70, TMX2, TPP1, TRAPPC12, TRAPPC6B, TRIP13, TRPM6, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TUBGCP2, UBR7, UFC1, UFM1, UNC80, USP18, VARS1, VLDLR, VPS11, VPS50, WARS2, WDR45B, WDR62, WDR73, YIF1B, YIPF5, ZNF335); SNV, CNV)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Autismus Tier 1 (ACSL4, ADNP, ADSL, AFF2, AHDC1, ALDH5A1, ALG12, ALG3, ALG6, ANK2, ANKRD17, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARID1A, ARID1B, ARID2, ARX, ASH1L, ASXL1, ASXL2, ASXL3, ATP13A2, ATP6AP2, ATP7A, ATRX, AUTS2, BCAP31, BPTF, BRD4, BRSK2, BRWD3, CACNG2, CAMTA1, CASK, CC2D1A, CCDC22, CDH15, CDK13, CDK16, CHD2, CHD3, CHD7, CHD8, CLCN4, CNKSR2, CNOT1, CRADD, CREBBP, CSDE1, CSNK2A1, CTCF, CTNNB1, CUL3, CUL4B, DKC1, DLG3, DLG4, DPF2, DSCAM, DYRK1A, EBP, EFTUD2, EHMT1, EIF2S3, EIF3F, EZH2, FGD1, FGF12, FLNA, FMR1, FOLR1, FOXP2, FRMPD4, FTSJ1, GATAD2B, GK, GNAI1, GPC3, GRIA3, GRIK2, GRIN2B, H1-4, HCFC1, HDAC8, HEPACAM, HNRNPK, HNRNPR, HNRNPU, HOXA1, HPRT1, HUWE1)	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	untersuchungstechnik
<p>Autismus Tier 1 (<i>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, QRIH1, 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</i>); SNV, CNV)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Autismus Tier 1 (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)	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Autismus Tier 1 (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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Marfan-Syndrom (FBN1, TGFB1, TGFB2, IPO8, COL1A2, FKBP14 ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Loeys-Dietz-Syndrom (COL3A1, IPO8, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2 ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Osteogenesis imperfecta Tier 1 ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Osteogenesis imperfecta Tier 2 ( <i>COPB2, MBTPS2, NUDT6, SGMS2, SUCO</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Joubert-Syndrom ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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, in-house pipeline
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, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Stäbchen-Zapfen Dystrophie ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Retinale Erkrankungen ( <i>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</i> )	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>Retinale Erkrankungen (FAM161A, TLCD3B, FLVCR1, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HCCS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT74, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIAA1549, KIF11, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MCOLN1, MED12, MERTK, MFRP, MFSD8, MIR204, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MTPP, MVK, MYO7A</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>
<p>Retinale Erkrankungen (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</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Retinale Erkrankungen (TIMM8A, TIMP3, TINF2, TMEM216, TMEM218, TMEM231, TMEM237, TOPORS, TPP1, TRAF3IP1, TREX1, TRNT1, TRPM1, TSPAN12, TTC8, TTL5, 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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Okulärer und okulokutaner Albinismus ( <i>GPR143, HPS1, HPS3, HPS4, HPS5, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Senior-Loken-Syndrom ( <i>CEP290, IQCB1, NPHP1, NPHP3, NPHP4, POC1B, SDCCAG8, TRAF3IP1, WDR19</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Kardiomyopathien (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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
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, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
DCM (ACTC1, MYH6, NKX2-5, ACTN2, BAG3, CDH2, DES, DMD, DOLK, DSC2, DSP, JUP, LAMP2, LMNA, MYBPC3, MYH7, NEXN, PLN, RBM20, SCN5A, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, EPG5, WWTR1, TBX20, ABCC9, GATA6, TAB2, TBX5, RAF1, ANK2, ANKRD1, CRYAB, FKRP, FKTN, FLII, JPH2, LDB3, MYLK3, MYPN, NRAP, PRDM16, RHBDF1, RPL3L, RRAGC, RRAGD, SCN1B, SGCD, SLC6A6, SPEG, TCAP, CSRP3, FLNC, MYL2, RYR2, TTR, CTF1, DTNA, EYA4, GATAD1, IDH2, ILK, LAMA4, NEBL, PLEKHM2, PSEN1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>Arrhythmien (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</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>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</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>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</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>Angeborene Herzfehler breit (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</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>
<p>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, TGFB1, TGFB2; CBS, FLCN, FOXE3, HCN4, MAT2A, MFAP5, SKI); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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, in-house pipeline
Long_QT (SCN5A, KCNQ1, CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNH2, KCNJ2, TRDN, ANK2, KCNE2, KCNJ5, SCN4B, SNTA1, TECRL); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
ARVC (FLNC, DES, DSC2, DSP, JUP, LMNA, TMEM43, PKP2, ANK2, CDH2, MYH7, PLN, SCN5A, TTN, TGFB3, MYL3, CAV3, CTNNA3, TJP1); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Meckel-Gruber-Syndrom ( <i>B9D1, B9D2, CC2D2A, CEP290, CSPP1, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TCTN3, TMEM107, TMEM216, TMEM231, TMEM67, TXNDC15, WDPCP</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Polyzystische Nierenerkrankung ( <i>ALG8, COL4A1, DNAJB11, DZIP1L, GANAB, GATM, HNF1B, LRP5, MUC1, NOTCH2, OFD1, PKD1, PKD2, PKHD1, PRKCSH, REN, SEC61A1, SEC63, TSC1, TSC2, TTC21B, UMOD, VHL</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Primäre Ziliäre Dyskinesie ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Sensorineurale Schwerhörigkeit ( <i>GJB2, GJB6</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Sensorineurale Schwerhörigkeit ( <i>ABHD12, ACTG1, ADGRV1, AIFM1, ALMS1, AP1S1, ATP1A3, ATP6V1B1, ATP6V1B2, BCS1L, BMP4, BSND, CABP2, CDC14A, CDC6, CDH23, CDT1, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLDN9, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A5, COL9A1, GSDME, PJVK, DHODH, DIAPH1, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, EIF4A3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FDXR, FGF10, FGF3, FGFR2, FGFR3, FOXI1, FOXI3, FRAS1, FREM2, GATA3, GGPS1, GIPC3, GJB2, GJB3, GJB6, GNAI3, GPSM2, GREB1L, GRHL2, GRIP1, GRXCR1, GSC, GSDME, HAAO, HMX1, HOXA2, HSD17B4, HSPA9, ILDR1</i> )	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
KARS1, KCNE1, KCNJ10, KCNJ16, KCNQ1, KCNQ4, KDM6A, KIT, KMT2D, LARS2, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MASP1, MITF, MN1, MPZL2, MSRB3, MT-RNR1, MT-TS1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OFD1, OGDHL, OPA1, ORC1, ORC4, ORC6, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTX2, P2RX2, PAX2, PAX3, PBX1, PCDH15, PDZD7, PJVK, PLCB4, PLS1, PNPT1, POLR1A, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RNF220, S1PR2, SALL1, SALL4, SERAC1, SERPINB6, SF3B4, SGPL1, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SOX10, SOX2, SPATA5, SPATA5L1, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Sensorineurale Schwerhörigkeit (USH1C, USH1G, USH2A, USP48, WFS1, WHRN, ABCC1, ACOX1, ATP11A, ATP2B2, BTB, CACNA1D, CCDC50, CEMIP, CLIC5, CLRN2, COG4, COL4A6, COL9A2, COL9A3, CRLS1, CRYM, DCDC2, DIABLO, DIAPH3, DMXL2, DNAJC3, ELMOD3, ESRP1, FOXF2, GDF6, GRXCR2, HARS2, HGF, HOMER2, KDM3B, KITLG, LETM1, MCM2, MET, MIR96, MORC2, NARS2, OXR1, PDSS1, PMP22, PIIP5K2, RIPOR2, ROR1, RPS28, SIX5, SLC26A5, SLC44A4, SNAI2, SPATC1L, SPNS2, SPTBN4, STX4, STXBP3, THOC1, TJP2, TMEM132E, TMTC2, TNC, TOP2B, TSPEAR, TUBB4B, WBP2, YARS1, MAN2B1, ANKH, ARSG, CDKN1C, EXOSC2, IARS2, MANBA, PEX1, PEX6, PEX26, PHYH, PSIP1, SGO2, TWNK, TYR); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline
Familiärer Brust- und Eierstockkrebs (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53, AKT1, APC, BLM, CDKN1B, CDKN2A, CTNNA1, DDB2, DICER1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FH, MAP2K1, MUTYH, NTHL1, POLD1, POLE, POLH, PTCH1, PTPN11, SLX4, SMARCA4, SMARCB1, SUFU, TOP3A, TRIM37, UBE2T, WRN, WT1, XPA, XPC, XRCC2); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Familiärer Brust- und Eierstockkrebs (Therapierelevanz) ( <i>BRCA1, BRCA2</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline
Li-Fraumeni-Syndrom ( <i>CHEK2, ACD, AKT1, ALK, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CTC1, DDB2, DICER1, DKC1, DOCK8, ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FH, FLCN, GATA1, GBA, GNAS, GPC3, GPR161, HRAS, KRAS, LIG4, MAD2L2, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NAF1, NBN, NF1, NF2, NHP2, NOP10, NRAS, NSD1, NTHL1</i>	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline
<i>PALB2, PARN, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD51C, RAD51D, RAF1, RB1, RECQL4, REST, RET, RIT1, RMRP, RNF43, RPA1, RPL11, RPL15, RPL23, RPL26, RPL31, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, STK11, STN1, SUFU, TERC, TERT, TINF2, TMEM127, TP53, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline
Lynch-Syndrom ( <i>MLH1, PMS2, MSH6, MSH2, EPCAM</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Polyposis (APC, BMPR1A, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, AXIN2, GREM1, EPCAM, MLH1, MSH2, MSH6, PMS2, EPCAM, MLH1, MSH2, MSH6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, SEC23B, SLX4, UBE2T, XRCC2, MSH3); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
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, in-house pipeline
Neurofibromatose / Schwannomatose (NF1, NF2, PRKAR1A, SMARCB1, LZTR1, SPRED1, GNAS, PTPN11, RAF1, RIT1, SOS1, EPCAM, MLH1, MSH2, MSH6, PMS2, BRAF, HOXB13, MAP2K1, PDGFRB, BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, KITLG, PALB2, SLX4); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Cowden Syndrom ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Gorlin-Goltz Syndrom ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Birt-Hogg-Dubé Syndrom ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Von-Hippel-Lindau Syndrom ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Multiple Exostosen ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Multiple endokrine Neoplasie ( <i>CDKN1B, MEN1, RET, CDC73, FH, MAX, NF1, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, UBE2T, TP53, AIP, CASR, DLST, DNMT3A, MDH2, SLC25A11, VHL</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Hereditäres Phäochromozytom-Paragangliom-Syndrom ( <i>DLST, EGLN1, EPAS1, FH, H3-3B, KIF1B, MAX, MDH2, MEN1, MERTK, MET, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SUCLG2, TMEM127, TP53, VHL</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	untersuchungstechnik
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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
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, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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, in-house pipeline
Blasen- und Harnleiterkrebs (EPCAM, MLH1, MSH2, MSH6, PMS2, BAP1, MUTYH, NTHL1, NF1, SMARCA4, SMARCB1, STK11, TP53, FH); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Endometriumkarzinom (FH, MLH1, MSH2, MSH6, PMS2, PTEN, EPCAM, MUTYH, NTHL1, POLD1, POLE, SMAD4); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
GIST (KIT, PDGFRA, SDHA, SDHB, SDHC, SDHD); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Magenkrebs (AAGAB, APC, ATM, BLM, BMPR1A, BRCA2, BUB1, BUB1B, BUB3, CASP10, CDH1, CDKN2A, CEP57, CHEK2, COL14A1, ERBB2, FGFR2, IL1B, IL1RN, IRF1, KLF6, KRAS, MDM2, MSH3, MUTYH, PIK3CA, POLD1, POLE, RPS20, SEMA4A, STK11, TP53, TRIP13, CTNNA1, EPCAM, MLH1, MSH2, MSH6, PDGFRA, PMS2, PTPN11, SMAD4); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
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, in-house pipeline
Schilddrüsenkarzinom (APC, CHEK2, CDC73, CDKN1B, DICER1, FLCN, POLH, PRKAR1A, PTEN, RET, TSC1, TSC2, WRN, TP53, XPA, XPC, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, MUTYH, GNAS, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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, in-house pipeline
Nierenkarzinom (APC, BAP1, CDC73, CDKN1B, CHEK2, DDB2, DICER1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FH, FLCN, MET, MLH1, MSH2, MSH6, NRAS, PMS2, POLH, PRKAR1A, PTEN, RAF1, SDHA, SDHAF2, SDHB, SDHC, SDHD, STK11, TMEM127, TP53, TSC1, TSC2, VHL, XPA, XPC); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
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, in-house pipeline
Melanom (BAP1, CDK4, CDKN2A, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, GBA, GNAS, MITE, POLH, POT1, PTEN, RB1, RECQL4, TERF2IP, WRN, XPA, XPC, ACD, BRCA2, TERT); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
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	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
<p>Tumordisposition breit (<i>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</i>); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik
Primäre Ovarialinsuffizienz Tier 1 ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Primäre Ovarialinsuffizienz Tier 2 ( <i>EIF2B4, GDF9, NOG, NUP107, POU5F1, RCBTB1, SOHLH2, BMPR1B, BUB1B, DACH2, EIF4ENIF1, FIGLA, LMNA, MSH5, NANOS3, PGRMC1, POF1B, POLR2C, POLR3H, PSMC3IP, SGO2, SYCP2L, KHDRBS1</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Primäre Ovarialinsuffizienz Tier 3 ( <i>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</i> ); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	untersuchungstechnik
<p>Erbliche Blutungerkrankungen (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, GF11B, 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; SNV, CNV VPS33B, VWF, WAS ); SNV, CNV</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>PCR-free Library Prep, Sequencing- by-synthesis, in-house pipeline</p>

Anlage zur Akkreditierungsurkunde D-ML-22580-01-00

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	untersuchungstechnik
Leukodystrophie (AARS1, AARS2, ABCD1, ALDH3A2, APP, ARSA, AUH, CLCN2, COL4A1, COL4A2, CSF1R, CTSB, CYP27A1, DARS1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, GALC, GBE1, GFAP, GJA1, GJB1, GJC2, GLA, GLB1, GSN, HEPACAM, HEXA, HMGCL, HTRA1, ITM2B, L2HGDH, LAMB1, LARS2, LIG3, LMNB1, MARS1, MCOLN1, MTHFR, NOTCH3, OCRL, PAH, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PLP1, POLR1C, POLR3A, POLR3B, PRNP, PSAP, PSEN1, PTEN, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RPS6KA3, SAMHD1, SNORD118, SPG11, SPG21, TREM2, TREX1, TTR, TUBB4A, TYMP, TYROBP, ZFYVE26, MLC1, CST3, CTC1, EARS2, GCDH, KIF5A, MAL, NPC1, PSEN2, TPP2); SNV, CNV	EDTA-Blut, DNA; DNA	PCR-free Library Prep, Sequencing-by-synthesis, in-house pipeline
Segregationsanalysen oder Untersuchung auf Anlageträgerschaften (z.B. <i>ABCC8, BRCA1, BRCA2, BRPF1, DARS2, DICER1, FIG4, MYBPC3, NCDN, NONO, PTPN11, TNNT2</i> )	EDTA-Blut, DNA; DNA	PCR und Sanger-Sequenzierung
Therapieindikation Olaparib (virtuelles Panel, Illumina Exome 2.5 and Mitochondrial Panel) ( <i>BRCA1, BRCA2</i> ); SNV	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by-synthesis, in-house pipeline
Sensorineurale Schwerhörigkeit (GJB2)	EDTA-Blut, DNA; DNA	PCR und Sanger-Sequenzierung