Liste der akkreditierten Verfahren (D-ML_13304-10 und D-ML_23124-01)

Gültig ab: **02.08.2024**

Inhaber der Akkreditierungsurkunde(n):

UKGM GmbH (Rudolf-Buchheim-Str. 8, 35392 Gießen) bzw. **MVZ UKGM GmbH** (Akkreditierungsurkunde noch ausstehend)

mit dem Standort

Zentrum für Humangenetik / MVZ Humangenetik

Baldingerstraße, 35043 Marburg

Untersuchungsgebiet:

Medizinische Laboratoriumsdiagnostik - 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.

Untersuchungsart:

Molekularbiologische Untersuchungen (Flex C) – Es handelt sich ausschließlich um *in Haus*-Verfahren.

Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenommen am** (erstellt am)
ACMG_Panel (v3.3); Gene: PMID 40568962	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-,CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (ThermoFisher)	**08.08.25** (23.07.25)
ALS (Amyotrophe Lateralsklerose); Gene: *ALS2, ANG, ANXA11, CHCHD10, CHMP2B, DCTN1, FIG4, FUS, HNRNP1, HNRNPA2B1, KIF5A, MAPT, MATR3, NEFH, NEK1, OPTN, PARK7, PFN1, PRPH, SETX, SIGMAR1, SLC52A2, SLC52A3, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TIA1, TRPM7, TUBA4A, UBQLN2, VAPB, VRK1*	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (21.12.23)
Arrhythmien, hereditäre; Gene: *ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DSG2, DSP, FLNC, GNB5, HCN4, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, PKP2, RYR2, SCN10A, SCN1B, SCN3B, SCN5A, SLC22A5, SLC4A3, SNTA1, TANGO2, TECRL, TGFB3, TMEM43, TRDN, TRPM4*	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (08.08.23)

Analyt (Messgröße)	Untersuchungs-material	Untersuchungstechnik	Anweisung (Version) Pipeline/Kit	Gerät	Im akkreditierten Bereich aufgenom-men am (erstellt am)
Ataxien (ohne Repeat-Erkrankungen); Gene: *AAAS, ABCB7, ABHD12, ADCY5, ADGRG1, AFG3L2, AMPD2, ANO10, AP1S2, APTX, ARMC9, ARSA, ATCAY, ATG7, ATM, ATP1A2, ATP1A3, ATP2B3, ATP7B, ATP8A2, AUH, B3GALNT2, BRF1, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CASK, CHMP1A, CHP1, CLCN2, CLN6, CLP1, COA7, COASY, COG5, COQ4, COQ8A, COQ8B, COX20, CP, CSTB, CWF19L1, CYP27A1, CYP2U1, DAB1, DAGLA, DARS2, DDHD2, DNAJC19, DNAJC5, DNMT1, DYNC1H1, EBF3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EPM2A, ERCC4, EXOSC3, EXOSC8, EXOSC9, FAT2, FGF14, FLVCR1, FOLR1, GALC, GBA2, GDAP2, GFAP, GJC2, GLRA1, GOSR2, GPAA1, GRID2, GRM1, GRN, HEXA, HEXB, IRF2BPL, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNQ3, KIF1C, LNPK, MARS2, MFN2, MMACHC, MORC2, MRE11, MSTO1, MTPAP, MTTP, MVK, NFASC, NHLRC1, NKX2-1, NKX6-2, NOP56, NPC1, NPC2, NUS1, OPA1, OPA3, OPHN1, PACS2, PAX6, PDYN, PEX16, PEX2, PEX6, PIK3R5, PITRM1, PLA2G6, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3K, POU4F1, PPP2R2B, PRDX3, PRICKLE1, PRKCG, PRNP, PRPS1, PRRT2, PTRH2, PUM1, RARS2, RNF170, RNF216, ROBO3, RORA, RUBCN, SACS, SAMD9L, SAR1B, SCN1A, SCN2A, SCN8A, SCYL1, SEPSECS, SETX, SIL1, SLC1A3, SLC25A46, SLC2A1, SLC39A8, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, SRD5A3, STUB1, SYNE1, SYNGAP1, TBC1D23, TDP1, TDP2, TGM6, THG1L, TMEM240, TOE1, TPP1, TSEN2, TSEN54, TTBK2, TTC19, TTPA, TUBA1A, TUBB2B, TUBB3, TUBB4A, TWNK, UBA5, UCHL1, VAMP1, VLDLR, VPS13D, VPS41, VPS53, VRK1, WDR73, WDR81, WFS1, WWOX, XRCC1*	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**22.01.24** (01.12.23)
Bannayan-Riley-Ruvalcaba-Syndrom, Gene: *AKT1, PIK3CA, PTEN, SEC23B*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1),Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (19.02.24)
Carney-Komplex, Gene: *PRKAR1A, STK11*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
CADASIL und Differenzialdiagnosen; Gene: *COL4A1, CTC1, GLA, HTRA1, NOTCH3, TREX1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (22.06.23)

Analyt (Messgröße)	Untersuchungs-material	Untersuchungstechnik	Anweisung (Version) Pipeline/Kit	Gerät	Im akkreditierten Bereich aufgenom-men am (erstellt am)
CMT/HMSN und Differenzialdiagnosen; Gene: *AARS1, ABCA1, ABHD12, AIFM1, ALS2, ANG, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CADM3, CCT5, CHCHD10, CNTNAP1, COA7, COX10, COX6A1, CTDP1, CYP27A1, DCAF8, DCTN1, DCTN2, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, FBLN5, FBXO38, FGD4, FIG4, FUS, FXN, GALC, GAN, GARS1, GBF1, GDAP1, GJB1, GLA, GNB4, GSN, HADHA, HADHB, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, HYCC1, IFRD1, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, KLHL13, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MTRFR, MYH14, NAGLU, NARS1, NDRG1, NDUFAF5, NEFH, NEFL, NGF, NIPA1, NTRK1, OPTN, PDHA1, PDK3, PEX12, PLEKHG5, PLP1, PMP2, PMP22, PNKP, POLG, POLR3B, PRDM12, PRPS1, PRX, PTRH2, RAB7A, REEP1, RETREG1, SACS, SARS1, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SCO2, SEPTIN9, SETX, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMAD3, SOD1, SORD, SPAST, SPG11, SPTLC1, SPTLC2, SURF1, SYT2, TARDBP, TECPR2, TNNT2, TRIM2, TRPA1, TRPV4, TTR, TUBB3, TYMP, VAPB, VCP, VWA1, WNK1, YARS1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (14.06.24)
Cowden-Syndrom, Gene: *AKT1, PIK3CA, PTEN, SEC23B*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1), Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Darmkrebs; Gene: *EPCAM (NM_002354.3), MLH1 (NM_000249.3), MSH2 (NM_000251.3), MSH6 (NM_000179.3), PMS2 (NM_000535.7), APC (NM_000038.6), BMPR1A (NM_004329.2), MUTYH (NM_001128425.2), NTHL1 (NM_002528.7), POLD1 (NM_001308632.1), POLE (NM_006231.4), PTEN (NM_001304717.5), SMAD4 (NM_005359.6), STK11 (NM_000455.5), CDH1 (NM_004360.5), CHEK2 (NM_007194.4)*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**24.09.24** (11.09.24)
Dystonie hereditär; Gene: *ADAR, ADCY5, AFG2B, AFG3L2, ANO3, AOPEP, APTX, ATM, ATP1A2, ATP1A3, ATP7B, BCPAP31, C19orf12, CACNA1A, CHMP2B, COASY, DCAF17, DDC, DLAT, DNAJC12, DRD2, ECHS1, EIF2AK2, FA2H, FASTKD2, FBXO7, FTL, GAMT, GCDH, GCH1, GNAL, GNAO1, HPCA, IRF2BPL, KCNMA1, KCTD17, KIF1C, KMT2B, MECR, NKX2-1, PANK2, PLA2G6, PRKN, PRKRA, PRRT2, SCN8A, SGCE, SLC30A10, SLC39A14, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VAC14, VPS13A, VPS16, YY1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1), Twist Exome 2.0, Bioinformatik-Pipeline:varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (14.12.23)
Ehlers-Danlos-Syndrom, klassischer Typ; Gene: *COL5A1, COL5A2, COL1A1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1), Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (08.10.20)

Analyt (Messgröße)	Untersuchungs-material	Untersuchungstechnik	Anweisung (Version) Pipeline/Kit	Gerät	Im akkreditierten Bereich aufgenom-men am (erstellt am)
Ehlers-Danlos-Syndrom, vaskulärer Typ; Gen: *COL3A1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1), Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (08.10.20)
Frontotemporale Demenz; Gene: *CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**17.09.25** (08.08.25)
Gliedergürtel-Muskeldystrophie/LGMD; Gene: *ABHD5, ACADVL, ACTA1, ACTN2, AGL, ANO5, ATP2A1, BAG3, BVES, CAPN3, CASQ1, CAV3, CHRND, CIAO1, CLCN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRPPA, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DOK7, DPM3, DYSF, EMD, ETFDH, FHL1, FKRP, FKTN, FLNC, GAA, GBE1, GFPT1, GMPPB, GNE, GYG1, HMGCR, HNRNPDL, LAMA2, LAMP2, LIMS2, LMNA, LPIN1, LRIF1, MATR3, MTM1, MYH14, MYH7, MYOT, NEB, ORAI1, PFKM, PGK1, PHKA1, PLEC, PNPLA2, POGLUT1, POLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PYGM, PYROXD1, RAPSN, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SMCHD1, STIM1, SYNE1, SYNE2, TCAP, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, VCP, VMA21*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0, Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**04.12.24** (26.11.24)
Hyperparathyreoidismus, primär; Gene: *CASR, CDC73, CDKN1B, MEN1, RET*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (24.11.21)
***Joubert-Syndrom und Differenzialdiagnosen*;** Gene: *ARL13B, ARL3, ARMC9, ATXN10, B9D1, B9D2, C2CD3, CELSR2, CEP41, CEP55, CPLANE1, CSPP1, EXOC8, HYLS1, IFT172, INPP5E, MKS1, NPHP3, PDE6D, PIBF1, POC1B, SUFU, TCTN1, TCTN3, TMEM107, TMEM138, TMEM237*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**29.05.25** (14.05.25)
Juvenile Makuladegeneration/M. Stargardt; Gene: *ABCA4, BEST1, C1QTNF5, CDH3, CFH, CLN3, CNGB3, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, FSCN2, IMPG1, IMPG2, IRX1, MFSD8, PROM1, PRPH2, RP1L1, RPGR, TIMP3, TTLL5*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**23.01.25** (18.12.24)

Analyt (Messgröße)	Untersuchungs-material	Untersuchungstechnik	Anweisung (Version) Pipeline/Kit	Gerät	Im akkreditierten Bereich aufgenom-men am (erstellt am)
Lynch-Syndrom (HNPCC), Gene: *MLH1, MSH2, MSH6, PMS2, EPCAM*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (08.10.20)
Kardiomyopathie (v2); Gene: *AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALMS1, ALPK3, ANK2, ANKRD1, ARSB, ATAD3A, BAG3, BCS1L, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CBL, CDH2, COA5, COA6, COA7, COX10, COX14, COX15, COX20, COX6A1, COX6B1, COX7B, CPT2, CRLS1, CRYAB, CSRP3, CYC1, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EPG5, ETFA, ETFDH, EYA4, FAH, FASTKD2, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, FNIP1, FOXRED1, GALNS, GATA6, GATAD1, GLA, GLB1, GNB5, GNS, GSN, GUSB, GYG1, HADHA, HADHB, HAMP, HCN4, HFE, HGSNAT, HJV, HRAS, IDH2, IDS, IDUA, JPH2, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, KRAS, LAMP2, LDB3, LMNA, LMOD2, LRPPRC, LYRM7, MAP2K1, MAP2K2, MIB1, MLYCD, MMACHC, MRAS, MRPL44, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYLK3, MYOZ2, MYPN, NAA15, NAGLU, NDUFA1, NDUFA10, NDUFA11, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB11, NDUFB3, NDUFB8, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEXN, NKX2-5, NONO, NRAP, NRAS, NUBPL, PCCA, PCCB, PDLIM3, PET100, PKP2, PLD1, PLN, PNPLA2, PPA2, PPCS, PPP1CB, PPP1R13L, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RASA2, RBM20, RHBDF1, RNF220, RPL3L, RPS6KB1, RRAGC, RRAGD, RYR2, SCN10A, SCN1B, SCN3B, SCN5A, SCO1, SCO2, SDHA, SDHAF1, SDHD, SGCD, SGSH, SHMT2, SHOC2, SLC22A5, SLC25A20, SLC25A4, SLC30A5, SLC40A1, SLC4A3, SLC6A6, SNTA1, SOS1, SOS2, SPEG, SPRED1, SPRED2, SURF1, TACO1, TAFAZZIN, TANGO2, TBX20, TBX5, TCAP, TECRL, TFR2, TMEM126B, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTC19, TTN, TTR, TULP3, UQCC2, UQCRB, VCL*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (19.03.21)
Katarakt, hereditär; Gene: *ABCB6, AGK, BCOR, BFSP1, BFSP2, CHMP4B, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, EPG5, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, FOXC1, FOXE3, FTL, FYCO1, GALK1, GCNT2, GEMIN4, GJA3, GJA8, GLA, HMX1, HSF4, HYCC1, JAM3, LEMD2, LIM2, LONP1, LSS, LTBP2, MAF, MIP, MYH9, NHS, OPA3, P3H2, PAX6, PITX2, PXDN, RAB18, RAB3GAP1, RAB3GAP2, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TMEM114, UNC45B, VIM, VSX2, WFS1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**15.07.25 (09.07.25)**

Analyt (Messgröße)	Untersuchungs-material	Untersuchungstechnik	Anweisung (Version) Pipeline/Kit	Gerät	Im akkreditierten Bereich aufgenom-men am (erstellt am)
Kraniosynostosen, hereditär; Gene: *ACTB, ACTG1, ADAMTSL4, AHDC1, ALPL, ALX4, ANKH, ARID1B, ARSB, ASXL1, ASXL3, AXIN2, B3GAT3, BCL11B, CDC45, CDK13, CHD5, COLEC11, CTSK, CYP26B1, DDX3X, DPF2, DPH1, EFNB1, ERF, ESCO2, FAM20C, FBX011, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FREM1, GLI3, GNAS, GNPTAB, GPC3, HNRNPK, HUWE1, IDS, IDUA, IFT122, IFT140, IHH, IL11RA, IL6ST, IRX5, JAG1, KANSL1, KAT6A, KAT6B, KPTN, LTBP1, MAN2B1, MASP1, MEGF8, MSX2, NFIA, NFIX, OGT, P4HB, PHEX, PJA1, POR, PPP1CB, PRRX1, PTCH1, RAB23, RECQL4, RNU12, RSPRY1, RUNX2, SEC24D, SIX1, SKI, SLC25A24, SMAD3, SMAD6, SMO, SOX6, SPECC1L, SPRY1, STAT3, TCF12, TFAP2B, TGFBR1, TGFBR2, TLK2, TMCO1, TRAF7, TWIST1, WDR35, ZEB2, ZIC1, ZNF462*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**17.09.24** (14.12.23)
Leukodystrophie und Leukoenzephalopathie; Gene: *AARS1, AARS2, ABCD1, ACBD5, ACO2, ACOX1, ADAR, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, BCAP31, CLCN2, CLDN11, CNP, CSF1R, CTC1, CYP27A1, DARS1, DARS2, DEGS1, EARS2, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, EXOSC8, FOLR1, FUCA1, GALC, GAN, GBE1, GCDH, GFAP, GJA1, GJC2, HEPACAM, HIKESHI, HSD17B4, HSPD1, HTRA1, HYCC1, IFIH1, ISCA2, KCNT1, KIF5A, L2HGDH, LAMA2, LAMB1, LMNB1, LSM11, MCOLN1, MLC1, NAXD, NAXE, NKX6-2, NOTCH3, OCLN, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHYH, PLAA, PLEKHG2, PLP1, POLR1A, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RAB11B, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU7-1, SAMHD1, SCP2, SLC16A2, SLC17A5, SLC25A12, SNORD118, SOX10, SPTAN1, STAT2, STN1, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYROBP, UFM1, VPS11, ZNHIT3*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**17.09.24** (01.02.24)
Magenkarzinom, diffuses, Gene: *CDH1, CTNNA1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (18.11.20)
Makuladystrophie; Gene: *ABCA4, BEST1, C1QTNF5, CDH3, CERKL, CFI, CHST6, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, FSCN2, GUCA1B, HMCN1, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RBP3, RBP4, RDH12, RDH5, RLBP1, RP1L1, RPGR, RPGRIP1, RS1, SIX6, TIMP3*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**11.12.24** (27.11.24)
Makrozephalie; Gene: *ABCC9, AKT1, AKT2, AKT3, AMER1, ASPA, BICD2, BLTP1, BRAF, BRWD3, CASK, CBL, CCND2, CDKN1C, CHD3, CHD4, CHD8, CUL4B, DHCR24, DICER1, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, FGFR3, GCDH, GFAP, GLI3, GNAQ, GPC3, GPC4, GPSM2, GRIA3, HEPACAM, HERC1, HRAS, HUWE1, INPPL1, ITCH, KDM1A, KIF7, KPTN, KRAS, L1CAM, LZTR1, MAP2K1, MAP2K2, MECP2, MED12, MLC1, MPDZ, MTOR, NF1, NFIA, NFIB, NFIX, NONO, NRAS, NSD1, OFD1, PIK3CA, PIK3R2, POLE, POLR3A, PPP1CB, PPP2R5D, PTCH1, PTEN, PTPN11, RAB39B, RAF1, RASA2, RHEB, RIN2, RIT1, RNF135, ROR2, RRAS, SEC23B, SETD2, SHANK3, SHOC2, SNX14, SOS1, SOS2,*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**17.09.24** (14.12.23)

SPRED1, STRADA, SUFU, SUZ12, SYN1, TBC1D7, TCF20, TMCO1, TMEM94, UPF3B, WNT5A, ZBTB20, ZDHHC9					
Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
Mamma- und Ovarialkarzinom (HBOC), 12 Gene: *ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, PALB2, RAD51C, RAD51D, PTEN, STK11, TP53*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**30.10.24** (06.09.24)
Mamma- und Ovarialkarzinom (HBOC), 6 Gene: *BRCA1, BRCA2, CHEK2, PALB2, RAD51C, ATM*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (04.01.24)
Mamma- und Ovarialkarzinom, Gene: *BRCA1, BRCA2*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (28.10.20)
Marfan-Syndrom und Loeys-Dietz-Syndrom, Gene: *FBN1, TGFBR1, TGFBR2*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (08.10.20)
Mikrozephalie, hereditär; Gene: *AARS1, ACTB, ADARB1, AFG2B, AGMO, AKT3, ANKLE2, AP4B1, AP4E1, AP4M1, AP4S1, ARCN1, ARPC4, ASPM, ATP1A2, ATP6V0A1, ATP6VOC, ATP9A, ATR, ATRX, BCL11A, BLM, BPTF, BRCA2, BRD4, BRIP1, BUB1B, CAMK2B, CASK, CCDC88A, CCND2, CDK5RAP2, CDT1, CENPF, CENPJ, CEP135, CEP152, CEP55, CEP57, CHAMP1, CHKA, CIT, CKAP2L, COASY, COPB1, CPSF3, CREBBP, CRIPT, CSNK2A1, CTCF, CTNNB1, CTSF, CTU2, DDX11, DHCR7, DIAPH1, DNA2, DNMT3A, DOHH, DONSON, DPM1, DPP6, DROSHA, DYNC1I2, DYRK1A, EFTUD2, EIF2S3, EIF5A, EP300, ERCC4, ERCC5, ERCC6, ERCC8, EXOC7, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FOXG1, GMNN, GPT2, GTF2E2, H4C3, HDAC8, HHAT, HMGB1, HPDL, IARS1, IER3IP1, IGF1, IGF1R, KIF11, KIF14, KIF1BP, KNL1, LAGE3, LARP7, LIG4, LMNB1, LMNB2, MCM7, MCPH1, MECP2, MED17, METTL5, MFSD2A, MINPP1, MORC2, MPLKIP, MRE11, MSMO1, MYCN, NAA20, NAPB, NARS, NBN, NCAPD2, NCAPD3, NDE1, NHEJ1, NIPBL, NSD2, NSRP1, NUP107, NUP188, NUP214, ORC1, ORC4, ORC6, OSGEP, PALB2, PCDH12, PCDHGC4, PCNT, PDCD6IP, PDHA1, PLK4, PNKP, POC1A, POGZ, PPIL1, PPP1R15B, PQBP1, PRIM1, PRUNE1, PTPN23, PUF60, PUS7, QARS1, RAD21, RAD50, RAD51, RAD51C, RBBP8, RMI1, RNU4ATAC, RPL10, RRP7A, RTTN, SARS1, SASS6, SLC1A4, SLC25A19, SLC38A3, SLC9A6, SLX4, SMC1A,*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (24.10.23)

SMC3, SMG8, STAMBP, STIL, SVBP, TAF13, TBX2, TBX4, TMX2, TNPO2, TOP3A, TP53RK, TRAIP, TRAPPC12, TRAPPC14, TRAPPC6B, TRAPPC9, TRIO, TRIP13, TRMT10A, TSEN15, TSEN54, TTC5, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, UBA5, UBE3A, UFC1, UFM1, UGP2, UNC80, VPS50, VRK1, WDR11, WDR37, WDR4, WDR62, WDR73, XRCC4, YIF1B, YIPF5, ZEB2, ZNF335, ZNF526, ZNF668					
Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
MODY, Gene: *ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1, RFX6*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24 (19.03.21)**
Multiple Endokrine Neoplasie Typ 1 /4, Gene: *CDC73, CDKN1B, MEN1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis); Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (25.08.20)
Multiple Endokrine Neoplasie Typ 2, Gene: *RET*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (25.08.20)
Myopathien, hereditär; Gene: *ABHD5, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGL, AK9, ALG14, ALG2, AMACR, AMPD1, ANO5, ASCC1, ASCC3, B3GALNT2, B4GAT1, BAG3, BCL11A, BICD2, BIN1, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CDH8, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL13A1, COL25A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A2, CPT1A, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DHX16, DMD, DNA2, DNAJB4, DNAJB6, DNM2, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DTNA, DYNC1H1, DYSF, ECEL1, EGR2, EMD, EPG5, ETFA, ETFB, ETFDH, EXOSC3, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GDAP1, GFER, GFPT1, GLA, GLRA1, GLRB, GMPPB, GNE, GYG1, HACD1, HADH, HADHA, HADHB, HINT1, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HNRNPU, IGHMBP2, ISCU, ITGA7, KANSL1, KBTBD13, KIF21A, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LAS1L, LDB3, LETM1, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4, MACF1, MAP3K20, MATR3, MEGF10, MFN2, MGME1, MICU1, MLIP, MPZ, MTM1, MTMR14, MTO1, MTRFR, MUSK, MYBPC1, MYBPC3, MYF5, MYH14, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYO18B, MYOD1, MYOT, MYPN, NEB, NEFL, OBSCN, OPA1, ORAI1, PAX7, PFKM, PGK1, PHOX2B, PIEZO2, PLEC, PNPLA2, POMGNT1, POMK, POMT1, POMT2, PPA2, PREPL, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RPH3A, RRM2B, RXYLT1, RYR1, RYR3, SBDS, SCN4A, SCO2, SECISBP2,*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**17.09.24** (22.11.23)

SELENON, SGCA, SGCB, SGCD, SGCE, SGCG, SGCZ, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A26, SLC25A3, SLC25A32, SLC25A4, SLC25A42, SLC5A7, SLC6A5, SMCHD1, SMN1, SNAP25, SPEG, SPG7, SPTBN4, SQSTM1, STAC3, STIM1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TCAP, TIA1, TK2, TMEM126B, TMEM43, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIP4, TRMT5, TRPV4, TTN, TUBB3, TWNK, TYMP, UBA1, UNC13A, UNC45B, VAMP1, VCP, VMA21, ZC4H2					
Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
Neuroendokrine Tumore / NET; Gene: *CDKN1B, MEN1, NF1, POT1, RET, TSC1, TSC2, VHL*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**14.11.24** (01.11.24)
Neurofibromatose Typ 1 / M. Recklinghausen: Gen: *NF1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Neurofibromatose Typ 2: Gen: *NF2*	EDTA-Blut, DNA; DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (02.12.20)
Neuropathie (Typ 1 und Typ 2), hereditär; Gene: *AAAS, AARS1, ABCA1, ABHD12, AGTPBP1, AGXT, AIFM1, ANG, AP1S1, APOA1, APTX, ARHGEF10, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CADM3, CCT5, CD59, CFAP276, CHCHD10, CNTNAP1, COA7, COX6A1, COX10, CPOX, CTDP1, CYP27A1, DARS2, DCAF8, DCTN1, DCTN2, DEGS1, DGAT2, DHTKD1, DNAJB2, DNAJB5, DNAJC3, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, ETFDH, FAH, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FUS, FXN, GAA, GALC, GAN, GARS1, GBA2, GBF1, GDAP1, GJB1, GJC2, GLA, GNB4, GSN, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HOXD10, HSPB1, HSPB3, HSPB8, HYCC1, IARS2, IFRD1, IGHMBP2, INF2, ITPR, JAG1, JPH1, KARS1, KCNA2, KIF1A, KIF1B, KIF5A, KLHL13, LITAF, LMNA, LRSAM1, LYST, MARS1, MCM3AP, MED25, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MTMR2, MTRFR, MTTP, MYH14, NAGA, NAGLU, NARS1, NDRG1, NDUFAF5, NEFH, NEFL, NGF, NHERF1, NIPA1, NTRK1, OPA1, OPA3, OPTN, PDHA1, PDK3, PDXK, PDYN, PEX10, PEX12, PEX7, PHYH, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PPOX, PRDM12, PRKCG, PRNP, PRPS1, PRX, PTEN, PTRH2, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN9A, SCO2, SCYL1, SEPTIN9, SETX, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMAD3, SMN1, SOD1, SORD, SOX10, SPAST,*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**17.09.24** (09.08.23)

SPG11, SPG7, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TARDBP, TDP1, TECPR2, TFG, TNNT2, TRIM2, TRPA1, TRPV4, TTPA, TTR, TUBB3, TWNK, TYMP, UBA5, VAPB, VCP, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, XRCC1, YARS1, ZFHX2, ZFYVE26					
Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
Nierenzellkarzinom, hereditär, Gene: *BAP1, FLCN, FH, MET, PTEN, SDHB, SDHD, TSC1, TSC2, VHL*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Noonan-Syndrom, Gene: *PTPN11, BRAF, KRAS, RAF1, RIT1, SOS1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (18.11.20)
Okuläre Fehlbildungen und Optikushypoplasie; Geme: *ABCB6, ACO2, AFG3L2, ALDH1A3, ALPK1, AP3B2, ATAD3A, ATG7, ATOH7, B3GALNT2, B3GLCT, BCOR, BLOC1S1, BMP4, BRAF, C19orf12, CAPN15, CASK, CHD7, CISD2, COL4A1, COL4A2, CYP1B1, DDHD2, DNAJC30, DNM1L, EPRS1, FAT1, FDXR, FOXC1, FOXE3, FRAS1, FREM1, FREM2, GDF3, GDF6, GRIP1, HCCS, HESX1, HIKESHI, HK1, HMX1, ISCA2, KANSL1, KIF7, KMT2D, KNSTRN, MAB21L2, MAG, MECR, MFF, MFN2, MFRP, MYRF, NBAS, NDUFA12, NR2F1, OPA1, OPA3, OTX2, PAX2, PAX6, PDSS1, PIGL, PIK3CD, PITX2, PITX3, PLK4, POLG, PRSS56, PUF60, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIX6, SLC25A46, SLC38A8, SLC44A1, SLC52A2, SMO, SMOC1, SOX2, SOX5, SPG7, SSBP1, STRA6, TBC1D20, TENM3, TFG, TIMM8A, TMEM126A, TMEM98, TUBA8, TUBGCP4, UBE3B, UCHL1, VAX1, VPS35L, VSX2, WFS1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**17.09.24** (20.06.23)
Okulärer und Okulokutaner Albinismus; Gene: *DCT, GPR143, LRMDA, OCA2, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**19.03.25** (14.03.25)
Oslersche Krankheit / Morbus Osler / Hereditäre hämorrhagische Teleangiektasie (HHT); *Gene: ACVRL1 (NM_000020.3), BMPR2 (NM_001204.7), ENG (NM_001114753.3), EPHB4 (NM_004444.5), GDF2 (NM_016204.4), RASA1 (NM_002890.3), SMAD4 (NM_005359.6), SOX18 (NM_018419.3)*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**20.09.24** (08.12.23)
Pankreaskarzinom, Gene: APC, ATM, *BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1,	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (10.11.20)

		Analyse; PCR, Sanger-Sequenzierung	Limbus, Rostock		
Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
Paragangliom-Phäochromozytom-Syndrom, Gene: *AIP, MAX, MEN1, NF1, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, TMEM127, VHL*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (17.11.20)
Paraplegie / Hereditäre Spastische Paraplegie (HSP); Gene: *ABCD1, ABHD12, ABHD16A, ACER3, ADAR, AFG2B, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMPD2, ANG, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATL1, ATP13A2, B4GALNT1, BCAS3, BICD2, BLOC1S1, BSCL2, C19orf12, CAPN1, CCDC82, CCT5, CDK16, CHP1, COQ4, CPT1C, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS1, DARS2, DDHD1, DDHD2, DDX3X, DNM2, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FBXO7, FIG4, GAD1, GALC, GAN, GBA2, GBE1, GCH1, GFAP, GJA1, GJC2, GLB1, GLRX5, GPT2, GRID2, HACE1, HEXA, HIKESHI, HPDL, HSPD1, IBA57, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, KPNA3, L1CAM, LYST, MAG, MAPK8IP3, MARS1, MARS2, MTPAP, MTRFR, NDUFA12, NIPA1, NKX6-2, NRCAM, NSRP1, NT5C2, OPA1, OPA3, PANK2, PCYT2, PDYN, PI4KA, PLA2G6, PLP1, PNPLA6, POLR3A, POLR3K, PSEN1, RAB3GAP2, REEP1, REEP2, RNASEH2B, RNF170, RTN2, SACS, SARS2, SELENOI, SERAC1, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SLC33A1, SOD1, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, TAF8, TARDBP, TECPR2, TFG, TH, TMEM63C, TNR, TUBB4A, UBAP1, UBQLN2, UCHL1, VAMP1, VAPB, VCP, VPS37A, WASHC5, WDR45B, WDR48, ZFYVE26, ZFYVE27*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (07.06.23)
Parkinson-Syndrom, hereditär; Gene: *ADH1C, ANO3, ATP13A2, ATP1A3, ATP6AP2, ATP7B, C19orf12, CHCHD2, CSF1R, DCTN1, DNAJC6, EIF4G1, FBXO7, FTL, GBA1, GCH1, GNAL, GRN, HTRA2, LRRK2, MAPT, NR4A2, PANK2, PARK7, PINK1, PLA2G6, PRKCG, PRKN, PRKRA, PSAP, PTRHD1, RAB39B, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SPG11, SPR, SYNJ1, TAF1, TH, TUBB4A, UCHL1, VPS13A, VPS13C, VPS35, WDR45, ZFYVE26*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.09.24** (10.05.23)
Peutz-Jeghers-Syndrom, Gen: *STK11*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Polyposis coli, Gene: *APC, BMPR1A, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (26.11.20)

		Sequenzierung			
Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
Porphyrie, hereditär; Gene: *ABCC2, ALAD, ALAS2, CDIN1, CLPX, CPOX, FECH, GATA1, HFE, HMBS, PPOX, SLC19A2, UROD, UROS*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**05.11.24** (25.09.45)
Prostatakarzinom: *ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, TP53*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (30.06.23)
RASopathie, Gene: *A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Rhabdoid-Prädispositions-Syndrom, Gene: *SMARCB1, SMARCA4*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Schilddrüsenkarzinom, medulläres, Gen: *RET*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (03.03.21)
Schilddrüsenkarzinom, papilläres, Gene: *APC, CDKN1B, DICER1, PRKAR1A, PTEN*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (03.02.23)
Schwannomatose, Gene: *LZTR1, NF2, SMARCB1*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)

Analyt (Messgröße)	Untersuchungs-material	Untersuchungstechnik	Anweisung (Version) Pipeline/Kit	Gerät	Im akkreditierten Bereich aufgenommen am (erstellt am)
Schwerhörigkeit, hereditär; Gene: *ABCC1, ABHD12, ACTG1, ADGRV1, AFG2A, AFG2B, AIFM1, ANKH, ARSG, ATOH1, ATP1A3, ATP11A, ATP2B2, ATP6V1B1, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, CLRN2, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, DPT, DSPP, DTNA, EDN3, EDNRB, ELMOD3, EPS8, EPHA10, EPS8L2, ERAL1, ESPN, ESRP1, ESRRB, EYA1, EYA4, FGF3, FGFR3, FITM2, FOXI1, GAB1, GAS2, GATA3, GDF6, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HARS2, HGF, HOMER2, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, KMT2D, LARS2, LHFPL5, LMX1A, LOXHD1, LRP5, LRTOMT, MAP1B, MARVELD2, MASP1, MCM2, MET, METTL13, MINAR2, MIR96, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, NARS2, NCOA3, NLRP3, NMNAT1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDE1C, PDZD7, PEX1, PEX26, PEX6, PHYH, PI4KB, PJVK, PLS1, PNPT1, POLR1B, POLR1C, POLR1D, POU3F4, POU4F3, PPIP5K2, PRKCB, PRPS1, PTPRQ, RAI1, RDX, REST, RIPOR2, ROR1, S1PR2, SALL1, SCD5, SERAC1, SERPINB6, SERPINF1, SIX1, SIX5, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC44A4, SLITRK6, SMPX, SNAI2, SOX10, SPATC1L, SPNS2, STRC, STX4, SYNE4, TBC1D24, TCOF1, TECTA, THOC1, TIMM8A, TJP2, TMC1, TMEM43, TMEM132E, TMEM145, TMIE, TMPRSS3, TMTC2, TNC, TOP2B, TPRN, TRIOBP, TRRAP, TSPEAR, TWNK, USH1C, USH1G, USH2A, USP48, WBP2, WFS1, WHRN*	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**07.10.24** (23.09.24)
Septo-optische Dysplasie; Gene: *ALG13, ARNT, FGF8, FGFR1, FLNA, GLI2, HESX1, OTX2, PAX6, PROKR2, PROP1, SOX2, SOX3, STAG2, TAX1BP3*	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**11.07.25** (11.07.25)
Spastische Paraplegie/SPG, hereditär; Gene: *ABCD1, ABHD12, ABHD16A, ACER3, ADAR, AFG2B, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMPD2, ANG, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATL1, ATP13A2, B4GALNT1, BCAS3, BICD2, BLOC1S1, BSCL2, C19orf12, CAPN1, CCDC82, CCT5, CDK16, CHP1, COQ4, CPT1C, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS1, DARS2, DDHD1, DDHD2, DDX3X, DNM2, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FBXO7, FIG4, GAD1, GALC, GAN, GBA2, GBE1, GCH1, GFAP, GJA1, GJC2, GLB1, GLRX5, GPT2, GRID2, HACE1, HEXA, HIKESHI, HPDL, HSPD1, IBA57, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, KPNA3, L1CAM, LYST, MAG, MAPK8IP3, MARS1, MARS2, MTPAP, MTRFR, NDUFA12, NIPA1, NKX6-2, NRCAM, NSRP1, NT5C2, OPA1, OPA3, PANK2, PCYT2, PDYN, PI4KA, PLA2G6, PLP1, PNPLA6, POLR3A, POLR3K, PSEN1, RAB3GAP2, REEP1, REEP2, RNASEH2B, RNF170, RTN2, SACS, SARS2, SELENOI, SERAC1, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SLC33A1, SOD1, SPART, SPAST, SPG11, SPG21, SPG7, SPR,*	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**20.09.24** (07.06.23)

SPTAN1, TAF8, TARDBP, TECPR2, TFG, TH, TMEM63C, TNR, TUBB4A, UBAP1, UBQLN2, UCHL1, VAMP1, VAPB, VCP, VPS37A, WASHC5, WDR45B, WDR48, ZFYVE26, ZFYVE27					
Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
Thorakale Aortenerweiterung, Gene: *ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (20.10.20)
Tuberöse Sklerose, Gene: *TSC1, TSC2*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Usher-Syndrom; Gene: *ABHD12 (NM_015600.5), ADGRV1 (NM_032119.4), ARSG (NM_014960.5), CDH23 (NM_022124.6), CEP250 NM_007186.6), CEP78 (NM_001098802.3), CIB2 (NM_006383.4, CLRN1 (NM_001195794.1), ESPN (NM_031475.3), MYO7A (NM_000260.4), PCDH15 (NM_001142763.2), PDZD7 (NM_001195263.2), PEX1 (NM_000466.3), PEX6 (NM_000287.4), TUBB4B (NM_006088.6), USH1C (NM_153676.4), USH1G (NM_173477.5), USH2A (NM_206933.4), WHRN (NM_015404.4)*	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**20.09.24** (15.09.23)
Von-Hippel-Lindau-Syndrom, Gen: *VHL*	EDTA-Blut, DNA	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)
Whole Exome-Sequenzierung (WES)	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**02.09.24** (22.02.23)
Wilms-Tumor: *WT1*	EDTA-Blut	Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina MiniSeq, SeqStudio (Thermo Fisher)	**02.09.24** (01.12.20)

Analyt (Messgröße)	Untersuchungs-material	Untersuchungstechnik	Anweisung (Version) Pipeline/Kit	Gerät	Im akkreditierten Bereich aufgenom-men am (erstellt am)
Ziliopathien, Gene: ADAMTS9, AGR2, AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BRWD1, C2CD3, CBY1, CC2D2A, CCDC39, CCDC40, CCNO, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP300, CFAP410, CFAP74, CILK1, CPLANE1, CRB2, CSPP1, CYS1, DAW1, DCDC2, DDX59, DHCR7, DLG5, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DRC2, DRC4, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, EXOC3L2, FAM149B1, FOXJ1, GAS2L2, GLI3, GLIS2, GRK2, HNF1B, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, IQCE, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, LAMA1, LBR, LRRC56, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, NEK1, NEK10, NEK8, NFKB1, NFKB2, NME5, NPHP1, NPHP3, NPHP4, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, PDIA6, PIBF1, PIK3C2A, PIK3CD, PIK3R1, PKD1, PKD2, PKHD1, PMM2, POC1B, PRKACA, PRKACB, PSKH1, RAG1, RAG2, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SBDS, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SPAG1, SPEF2, STK36, SUFU, TBC1D32, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TTC12, TTC21B, TTC8, TUBB4B, TXNDC15, VPS13B, WDPCP, WDR19, WDR35, WFDC2, XPNPEP3, ZMYND10, ZNF423, ZSWIM6	EDTA-Blut, DNA	Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung	AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock	Illumina NextSeq, SeqStudio (Thermo Fisher)	**10.03.25** (17.02.25)

Analyt (Messgröße)	**Untersuchungs-material**	**Untersuchungstechnik**	**Anweisung (Version) Pipeline/Kit**	**Gerät**	**Im akkreditierten Bereich aufgenom-men am** (erstellt am)
ABCC6-Defizienz, Gen: ABCC6	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Apr 2024)
Caveolin-Defizienz, Gen: CAV3	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Apr 2024)
CMT1, Gen: PMP22	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**24.01.25** (Dez 2024)
Fumarat-Hydratase-Defizienz, Gen: FH	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Mai. 2024)
HBOC / Mamma- und Ovarialkarzinom, Gene: *BRCA1, BRCA2*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Okt 2024)
HBOC / Mamma- und Ovarialkarzinom (HBOC), Gene: *BRCA1, BRCA2, CHEK2, PALB2, RAD51C*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Juli 2024)
Hypercholesterinämie, hereditär; Gen: LDLR	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo F isher)	**14.01.25** (Dez 2024)
Juvenile Polyposis / JPS; Gene: BMPR1A, PTEN, SMAD4	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Aug 2024)
Langes QT-Syndrom; Gene: KCNQ1, KCNH2, KCNE1, KCNE2, KCNJ2	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Apr. 2024)
Lynch-Syndrom, Gene: MLH1, MSH2, MSH6, PMS2, EPCAM	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Sept 2024)
Multiple Endokrine Neoplasie Typ 1, Gen: *MEN1*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**14.01.25** (Dez 2024)
Polyposis coli, Gene: APC, MUTYH	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Feb 2024)
Porphyrie; Gene: ALAD, CPOX, FECH, HMBS, PPOX, UROS, UROD	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**10.12.24** (Nov 2024)
Peutz-Jeghers-Syndrom, Gen: *STK11*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Juli 2024)
SHOX-Ausfall, Gen: SHOX	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Apr. 2024)
Tuberöse Sklerose, Gene: *TSC1, TSC2*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**06.11.24** (Okt 2024)
Marfan-Syndrom und Loeys-Dietz-Syndrom, Gene: *FBN1, TGFBR1, TGFBR2*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**02.09.24** (Jan 2024)
Neurofibromatose Typ 1, Gen: *NF1*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**06.11.24** (Okt 2024)
Neurofibromatose Typ 1, Gen: *NF2*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**06.11.24** (Okt 2024)
Ehlers-Danlos-Syndrom, vaskulärer Typ, Gen: *COL3A1*	EDTA-Blut, DNA	MLPA	AA_MLPA (v2)	SeqStudio (Thermo Fisher)	**17.12.24** (Dez 2024)