

## Liste der akkreditierten Verfahren (D-ML\_13304-10 und D-ML\_23124-01)

Gültig ab:

Inhaber der Akkreditierungsurkunden:

**UKGM GmbH:** seit 02.08.24 (Rudolf-Buchheim Str. 8, 35392 Gießen) und **MVZ UKGM GmbH:** angestrebt in 2025

(Baldingerstr., 35043 Marburg)

mit dem Standort

**Zentrum für Humangenetik und 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-<br>material | Untersuchungstechnik                                                                                                                                 | Anweisung (Version)<br>Pipeline/Kit                                                                                       | Gerät                                      | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
|------------------------------------------------------------------------------------------------------|----------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|-----------------------------------------------------------|
| <b>Absence-Epilepsie;</b> Gene: <i>CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, KCNMA1, SLC12A5, SLC2A1</i> | 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) | Dez 2023                                                  |
| <b>ACMG_Panel</b> (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) | Jul 2025                                                  |
| <b>Alport-Syndrom;</b> Gene: <i>COL4A5, COL4A3, COL4A4, FN1, MYH9</i>                                | 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) | Okt 2023                                                  |

## Liste

| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Untersuchungs-material | Untersuchungstechnik                                                                                                                                 | Anweisung (Version) Pipeline/Kit                                                                                          | Gerät                                       | Erstellt/im akkreditierten Bereich aufgenommen am |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------|---------------------------------------------|---------------------------------------------------|
| <b>ALS (Amyotrophe Lateralsklerose);</b> Gene: <i>ALS2, ANG, ANXA11, CHCHD10, CHMP2B, DCTN1, FIG4, FUS, HNRNP1, HNRNPAB1, KIF5A, MAPT, MATR3, NEFH, NEK1, OPTN, PARK7, PFN1, PRPH, SETX, SIGMAR1, SLC52A2, SLC52A3, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TIA1, TRPM7, TUBA4A, UBQLN2, VAPB, VRK1</i>                                                                                                                                                                                                                                                                                                                                           | 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) | Dez 2023                                          |
| <b>Alzheimer-Demenz;</b> Gene: <i>APOE, APP, CHMP2B, CSF1R, GRN, PSEN1, PSEN2, SQSTM1, TBK1, TREM2, VCP</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |                        | 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) | Jan 2025                                          |
| <b>Amyloidose, hereditär;</b> Gene: <i>APOA1, APOA2, APOC2, APOC3, B2M, CST3, FGA, GSN, LYZ, MEFV, NLRP3, OSMR, TTR</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | 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) | Jan 2025                                          |
| <b>Angelman-Syndrom und Differenzialdiagnosen;</b> Gene: <i>ATRX, CDKL5, CNTNAP2, EHMT1, FOXG1, KMT2C, MECP2, NRXN1, SLC9A6, STXBP1, TCF4, UBE3A</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | 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) | Jan 2025                                          |
| <b>Anophthalmie, Mikrophthalmie;</b> Gene: <i>ALDH1A3, BCOR, BMP4, CAPN15, COL4A1, FAT1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, GRIP1, HCCS, KMT2D, MAB21L2, MFRP, MYRF, OTX2, PAX2, PAX6, PITX2, PLK4, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, SHH, SIX6, SMO, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TMEM98, TUBGCP4, VPS35L, VSX2</i>                                                                                                                                                                                                                                                                                                      | 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) | Okt 2023                                          |
| <b>Arrhythmien, hereditäre;</b> Gene: <i>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</i>                                                                                                                                                                                                                                                                                                                         | 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) | Aug 23                                            |
| <b>Arthrogrypose, hereditär;</b> Gene: <i>ACADM, ACADS, ACADVL, ACTA1, ACTB, ACTG1, ADAMTS10, ADAMTS15, ADCY6, ADGRG6, ADSL, AGL, AGRN, AIMP1, AK9, ALG14, ALG2, ALG3, AMER1, ANOS, ANTXR2, ASCC1, ASXL1, ATAD1, ATP1A2, ATP1A3, B3GALT2, B4GAT1, BAG3, BICD2, BIN1, BLTP1, CACNA1E, CACNB2, CAPN3, CAV3, CCDC78, CFL2, CHAT, CHD8, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHUK, CNTN1, CNTNAP1, COASY, COL12A1, COL13A1, COL25A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRLF1, CRPPA, CRYAB, CTDPI, CUL4B, DAG1, DENND5A, DES, DHCR24, DMD, DNABJ6, DNM2, DOK7, DOLK, DPAGT1, DPM2, DYNC1H1, DYNC2H1, DYSF, EBP, ECEL1, EGR2,</i> | 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) | Aug 23                                            |

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| EMD, ERBB3, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC8, ERGIC1, ETFA, ETFB, ETFDH, EXOSC3, FAM20C, FBN2, FGFR2, FGFR3, FHL1, FILIP1, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FLNC, FLVCR2, FOXP3, GAA, GARS1, GBA1, GBE1, GDAP1, GFPT1, GLDN, GLE1, GMPPB, GNE, HADH, HADHA, HADHB, HNRNPH1, HSPG2, IBA57, IGHMBP2, IRF6, ISCU, ITGA7, KAT6B, KBTBD13, KCNK3, KIDINS220, KIF21A, KLHL40, KLHL41, KLHL7, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LAS1L, LDB3, LGI4, LMNA, LMOD3, LMX1B, LPIN1, LRP4, MACF1, MAGEL2, MATR3, MED12, MEGF10, MFN2, MICU1, MPZ, MTM1, MTMR1, MTMR14, MUSK, MYBPC1, MYF6, MYH2, MYH3, MYH7, MYH8, MYL11, MYMK, MYO9A, MYOD1, MYOT, NALCN, NEB, NEFL, NEK9, NUP88, ORAI1, PDHA1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PHGDH, PIEZO2, PIGH, PIP5K1C, PLEC, PLOD1, PLOD2, PNPLA2, POLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PREPL, PRG4, PYGM, RAC1, RASPN, RARS2, RHEB, RIPK4, RPH3A, RRM2B, RXYLT1, RYR1, SCARF2, SCN1A, SCN4A, SCYL2, SELENON, SEPSECS, SGCA, SGCB, SGCD, SGCG, SIL1, SKI, SLC18A3, SLC22A5, SLC25A1, SLC25A26, SLC29A3, SLC35A3, SLC52A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMN1, SMPD4, SNAP25, SPEG, STAC3, STIM1, SVIL, SYNE1, SYT2, TBCD, TCAP, TGF2, TGF3, TGFBR1, TGFBR2, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TOR1A, TOR1AIP1, TPM2, TPM3, TRIM32, TRIP4, TRPV4, TSEN2, TSEN34, TSEN54, TTN, TUBB2A, TUBB2B, UBA1, UNC13A, VAMP1, VIPAS39, VMA21, VPS33B, ZBTB33, ZBTB42, ZC4H2, ZMPSTE24 |                |                                                                                                                                                      |                                                                                                                           |                                             |          |
| <b>ARVD</b> ; Gene: BAG3, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LEMD2, LMNA, MYBPC3, MYH7, MYL3, NKX2-5, PKP2, PLN, RYR2, SCN5A, TGF3, TJP1, TMEM43, TTN                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | 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) | Dez 2024 |
| <b>Ataxien (ohne Repeat-Erkrankungen)</b> ; 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, FXN, GALT, GBA2, GDAP2, GFAP, GJC2, GLRA1, GOSR2, GPAA1, GRID2, GRM1, GRN, HEXA, HEXB, IRF2BPL, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNQ3, KIF1C, LNPB, MARS2, MFN2, MMACHC, MORC2, MRE11, MSTO1, MTPAP, MTPP, MVK, NFASC, NHLRC1, NKX2-1, NKX2-2, 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,                                                                                                                                                                                                                                                                                                                                                                  | 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) | Dez 2023 |

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| 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                                                                                                                                                                                                                                                                                    |                            |                                                                                                                                                     |                                                                                                                                          |                                                   |                                                           |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------|-----------------------------------------------------------|
| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                      | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
| <b>Bannayan-Riley-Ruvalcaba-Syndrom</b> , Gene: <i>AKT1, PIK3CA, PTEN, SEC23B</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1), Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Bartter-Syndrom</b> ; Gene: <i>ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, GNA11, HSD11B2, INSR, KCNJ1, KCNK10, KLHL3, MAGED2, NR3C2, SCNN1A, SCNN1G, SLC12A1, SLC12A2, SLC12A3, SLC4A1, SLC4A4, TRPM6, WNK1, WNK4</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jul 25                                                    |
| <b>Bindegeweserkrankungen</b> , Gene: <i>ACTA2, ADAMTS10, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ARIH1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A3, COL4A5, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE1, FOXE3, GATA5, GORAB, HRAS, JAG1, JAM3, LOX, LOXL3, LTBP1, LTBP3, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NAA10, NOTCH1, PHYKPL, PLOD1, PLOD3, PRDM5, PRKG1, PTSS1, PYCR1, RIN2, ROBO4, RPS6KA3, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB1, TGFB2, TGFB3, TGFB1R1, TGFB2R2, TNXB, TWIST2, UPF3B, ZDHHC9, ZNF469</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jul 24                                                    |
| <b>Blepharophimose-Ptoxis, Epicanthus inversus-Syndrom und Differenzialdiagnosen</b> ; Gene: <i>BRAF, DHCR7, FOXL2, KANSL1, KRAS, LZTR1, MAP2K1, MASP1, NRAS, PTPN11, RAF1, RIT1, SOS1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 24                                                    |
| <b>Carney-Komplex</b> , Gene: <i>PRKAR1A, STK11</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                    | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                      | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
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| <b>CADASIL und Differenzialdiagnosen;</b> Gene: <i>COL4A1, CTC1, GLA, HTRA1, NOTCH3, TREX1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jun 24                                                    |
| <b>Cholangiokarzinom;</b> Gene: <i>APC, BRCA1, BRCA2, EPCAM, MLH1, MSH2, MSH6, PMS2</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Okt 23                                                    |
| <b>Cholestase;</b> Gene: <i>ABCB11, ABCB4, ABCC2, ACOX2, ADK, AKR1D1, ALDOB, AMACR, ASS1, ATP7B, ATP8B1, BAAT, BCS1L, CC2D2A, CFTR, CLDN1, COG7, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DHCR7, EPHX1, FAH, GALE, GALM, GALT, GBA1, GBE1, GNAS, HADHA, HFE, HNF1B, HSD3B7, IARS1, INVS, JAG1, KIF12, LIPA, LSR, MKS1, MMP15, MPI, MPV17, MVK, MYO5B, NBAS, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PKHD1, POLG, RINT1, SEMA7A, SERPINA1, SLC25A13, SLC27A5, SLC51A, SLCO1B1, SLCO1B3, SMPD1, TALDO1, TJP2, TMEM216, TRMU, UGT1A1, UNC45A, USP53, UTP4, VIPAS39, VPS33B, VPS50, YARS1, ZFYVE19</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                            | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 24                                                    |
| <b>CMT/HMSN und Differenzialdiagnosen;</b> Gene: <i>AARS1, ABCA1, ABHD12, AIFM1, ALS2, ANG, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSC12, 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</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jun 24                                                    |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                              | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                       | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------|-----------------------------------------------------------|
| <b>CMT2/HMSNII und Differenzialdiagnosen;</b> Gene: <i>AARS1, AIFM1, ATP1A1, BSCL2, CHCHD10, COX6A1, DCAF8, DGAT2, DHTKD1, DNAJB2, DNM2, DRP2, DYNC1H1, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, KIF5A, LMNA, LRSAM1, MARS1, MCM3AP, MFN2, MME, MORC2, MPV17, MPZ, MTRFR, NAGLU, NEFH, NEFL, PDK3, PLEKHG5, PMP22, PNKP, PRPS1, RAB7A, SACS, SLC25A46, SORD, SPG11, TFG, TRIM2, TRPV4, TTR, TUBB3, VCP, YARS1</i>                | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1), Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jan 25                                                    |
| <b>Cowden-Syndrom,</b> Gene: <i>AKT1, PIK3CA, PTEN, SEC23B</i>                                                                                                                                                                                                                                                                                                                                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1), Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Darmkrebs;</b> Gene: <i>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)</i>                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1), Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Sep 24                                                    |
| <b>Dyskeratosis congenita;</b> Gene: <i>ACD, AANAPC1, ATP2A2, ATP2C1, CAST, CDSN, CHST8, CSTA, CTC1, DCLRE1B, DKC1, DSG1, DSP, FERMT1, FLG2, GRHL2, LIG4, MBTPS2, NAF1, NHEJ1, NHP2, NOP10, NPM1, PARN, POT1, RECQL4, RTEL1, SERPINB8, TERC, TERT, TGM5, TINF2, TRPV3, USB1, WRAP53</i>                                                                                                                                                                                         | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1), Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jun 24                                                    |
| <b>Dystonie hereditär;</b> Gene: <i>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, MECP, NKX2-1, PANK2, PLA2G6, PRKN, PRKRA, PRRT2, SCN8A, SGCE, SLC30A10, SLC39A14, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VAC14, VPS13A, VPS16, YY1</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1), Twist Exome 2.0,<br>Bioinformatik-<br>Pipeline:varfeed 2.5.1,<br>Limbus, Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 23                                                    |
| <b>Ehlers-Danlos-Syndrom, klassischer Typ;</b> Gene: <i>COL5A1, COL5A2, COL1A1</i>                                                                                                                                                                                                                                                                                                                                                                                              | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1), Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                    | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Okt 20                                                    |
| <b>Ehlers-Danlos-Syndrom, vaskulärer Typ;</b> Gen: <i>COL3A1</i>                                                                                                                                                                                                                                                                                                                                                                                                                | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1), Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                    | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Okt 20                                                    |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                               | Anweisung (Version)<br>Pipeline/Kit                                                                                        | Gerät                                       | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
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| <b>Entwicklungsstörung und geistige Behinderung;</b> Gene: A2M, AAAS, AARS1, AASS, ABCC8, ABCD1, ABHD5, ACADM, ACAT1, ACO2, ACOX1, ACSL4, ACTB, ACTG1, ACY1, ADAMTSL2, ADAR, ADNP, ADSL, AFF2, AFG2A, AGA, AGTR2, AHDC1, AHI1, AIFM1, AIMP1, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG12, ALG13, ALG2, ALG3, ALG6, ALMS1, ALX4, AMER1, AMT, ANK3, ANKRD11, AP1S1, AP1S2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, APTX, AR, ARFGF2, ARG1, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARL6, ARSA, ARSL, ARX, ASAH1, ASL, ASNS, ASPM, ASS1, ASXL1, ASXL3, ATIC, ATL1, ATP10A, ATP13A2, ATP1A2, ATP1A3, ATP2A2, ATP6AP2, ATP6VOA2, ATP7A, ATRX, AUH, AUTS2, AVPR1A, AVPR2, B4GALT1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCL11A, BCOR, BCS1L, BDNF, BICRA, BIN1, BLM, BRAF, BRAT1, BRD4, BRSK2, BRWD3, BSCL2, BSND, BTB, BUB1B, C12orf57, CA2, CA8, CACNA1A, CACNA1C, CACNA1E, CACNG2, CAMK2B, CAMTA1, CANT1, CASK, CBL, CBS, CC2D1A, CC2D2A, CCDC22, CCDC88C, CDC42, CDH15, CDK13, CDK16, CDK5RAP2, CDKL5, CDKN1C, CENPJ, CEP152, CEP290, CEP41, CEP57, CERT1, CFH, CHAMP1, CHD2, CHD7, CHD8, CHKB, CHRNA4, CHST14, CIC, CLCN4, CLIC2, CLN3, CLN5, CLN6, CLN8, CLTC, CNBP, CNKSR2, CNNM2, CNTNAP2, CNTNAP5, COA5, COG5, COG7, COG8, COL18A1, COL4A1, COQ2, COQ8A, COQ9, COX15, COX6B1, CP, CPA6, CPANE1, CPS1, CRADD, CRBN, CREBBP, CRLF1, CRPPA, CSNK2A1, CSTB, CTC1, CTCF, CTD1P1, CTNNA1, CTNND2, CTSB, CTSD, CTSF, CUL3, CUL4B, CYB5R3, CYP27A1, D2HGDH, DAG1, DARS2, DBT, DCAF17, DCX, DDC, DDX3X, DEAF1, DHCR24, DHCR7, DKC1, DLD, DLG3, DLGAP2, DMD, DNAJC19, DNM1, DNM1L, DNMT3A, DNMT3B, DOCK4, DOCK6, DPAGT1, DPF2, DPM1, DPP10, DPP6, DPYD, DPYS, DUOX2, DYNC1H1, DYRK1A, EBF3, EBP, EEF1A2, EFN1, EFTUD2, EHMT1, EIF2AK3, EIF2B1, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELOVL4, EP300, EPM2A, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN2, ESCO2, ETHE1, EVC, EVC2, EXOSC3, EZH2, FA2H, FAAH2, FAF1, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCG, FANCL, FARS2, FASTKD2, FBN1, FBXO11, FGD1, FGF1, FGF14, FGFR1, FGFR2, FGFR3, FH, FKR1, FKTN, FLNA, FLNB, FMN2, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FOXRED1, FRAS1, FREM2, FRMPD4, FTO, FTSJ1, FUCA1, G6PC3, GABBR2, GABRA1, GABRB3, GABRG2, GALT, GALE, GALT, GAMT, GAN, GATAD2B, GATM, GBA1, GBE1, GCH1, GCK, GCSH, GDI1, GFAP, GFM1, GHR, GJB1, GJC2, GK, GLB1, GLI3, GLRA1, GLUL, GLYCTK, GM2A, GNAO1, GNAS, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GPI, GRIA3, GRIK2, GRIK3, GRIN1, GRIN2A, GRIN2B, GRIP1, GRM1, GRN, GSPT2, GSS, GTF2H5, GUSB, HADH, HAX1, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, HEXA, HEXB, HGSNAT, HIVEP2, HLCS, HNRNP1, HNRNP1, HOXA1, HPD, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HSPG2, HUWE1, HYCC1, IDS, IDUA, IER3IP1, IGBP1, IGF1, IGF1R, IKBKG, IL1RAPL1, IMPA1, INSR, IQSEC2, IRX5, ITGA7, ITPR1, IVD, JAG1, JAM3, KANSL1, KAT6A, KAT6B, KATNAL2, KCNA2, KCNB1, KCNC3, KCNH1, KCNJ1, KCNJ10, KCNJ11, KCNK9, KCNQ2, KCNQ3, KCNT1, KCTD13, KCTD7, KDM5B, KDM5C, KDM6A, KIF11, KIF1A, KIF21A, KIF5A, KIF7, KIRREL3, KLF8, KMT2A, KMT2B, KMT2D, KMT2E, KMT5B, KRAS, L1CAM, L2HGDH, LAMA2, LAMC3, LAMP2, LARGE1, | 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) | Mrz 25                                                    |

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| <p>LAS1L, LBR, LHX3, LIG4, LMBRD1, LMNB1, LRP2, LRPPRC, LYST, LZTR1, MACF1, MAF, MAGEL2, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MAP2K1, MAP2K2, MAPK8IP3, MAT1A, MBD5, MBTPS2, MCCC1, MCCC2, MCOLN1, MCPH1, MECP2, MED12, MED13L, MED17, MED23, MEF2C, MFSD8, MGAT2, MID1, MKKS, MLC1, MLYCD, MMAA, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MPV17, MRAP, MTFMT, MTHFR, MTOR, MTR, MVK, MYCN, MYH3, MYO5A, MYT1L, NAA10, NAA15, NAGA, NAGLU, NAGS, NALCN, NBN, NDE1, NDN, NDP, NDST1, NDUFA1, NDUFA2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NEU1, NEXMIF, NF1, NFIX, NGF, NGLY1, NHEJ1, NHLRC1, NHP2, NHS, NIPBL, NKX2-1, NKX2-5, NLGN1, NLGN3, NLGN4X, NONO, NPC1, NPC2, NPHP1, NPHP3, NR2F1, NRAS, NRXN1, NSD1, NSDHL, NSUN2, NTNG1, NTRK1, NUBPL, NUP62, OCLN, OCRL, OFD1, OGT, OPA3, OPHN1, ORC1, OTC, OTX2, PACS1, PACS2, PAFAH1B1, PAH, PAK3, PAX3, PAX6, PC, PCBD1, PCDH19, PCGF2, PCNT, PDE4D, PDHA1, PDHX, PDP1, PDSS1, PDSS2, PDYN, PEPD, PEX1, PEX10, PEX13, PEX19, PEX2, PEX3, PEX5, PEX7, PGAP3, PGK1, PHF21A, PHF6, PHF8, PHGDH, PHIP, PHKA2, PHKG2, PIGA, PIGL, PIGN, PIGO, PIGV, PIK3R2, PIP5K1B, PLA2G6, PLCB1, PLOD1, PLP1, PMM2, PNKP, PNP, POGZ, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMT1, POMT2, PON3, PORCN, POU1F1, PPM1D, PPOX, PPP1CB, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRKAR1A, PRKCG, PRMT7, PRODH, PRPS1, PRSS12, PSAP, PSPH, PTCH1, PTCHD1, PTEN, PTPN11, PTS, PURA, PUS1, PYCR1, PYGL, QDPR, RAB18, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAD21, RAI1, RAPS1, RARS2, RBBP8, RBFOX1, RBM10, RELN, RFT1, RIT1, RNASEH2A, RNASEH2B, RNASEH2C, RNU4-2, ROGDI, RORA, RPGRIP1L, RPL10, RPS6KA3, RRM2B, SACS, SALL1, SAMHD1, SATB2, SBDS, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO2, SDCCAG8, SEMA3E, SETBP1, SETD5, SGSH, SHANK2, SHANK3, SHOC2, SHROOM4, SIL1, SIN3A, SIX3, SKI, SKIC3, SLC12A1, SLC13A5, SLC16A2, SLC17A5, SLC20A2, SLC25A12, SLC25A13, SLC25A15, SLC25A2, SLC25A22, SLC2A1, SLC2A2, SLC35C1, SLC3A1, SLC46A1, SLC4A4, SLC6A1, SLC6A3, SLC6A4, SLC6A8, SLC7A7, SLC9A6, SLC9A9, SLX4, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMOG1, SMPD1, SMS, SNAP25, SNAP29, SNIP1, SNRPN, SOBP, SON, SOS1, SOS2, SOX10, SOX11, SOX2, SOX3, SOX4, SOX5, SPART, SPAST, SPG11, SPINK5, SPR, SPRED1, SPTAN1, SRCAP, SRD5A3, SRPX2, ST3GAL3, ST3GAL5, STAG1, STAG2, STAT5B, STRA6, STX11, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYP, SYT14, TAF1, TAF6, TAT, TBC1D24, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TCF20, TCF4, TCN2, TECR, TEO2, TET3, TG, TGFB1, TGFB2, TGIF1, TH, THOC2, THRB, TIMM8A, TINF2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TPH2, TPK1, TPP1, TRAPPC9, TREX1, TRHR, TRIM32, TRIO, TRIP12, TRRAP, TSC1, TSC2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TTC19, TTC8, TTI2, TUBA1A, TUBA8, TUBB2B, TUBB3, TUBGCP6, TUSC3, TWIST1, TWNK, UBE2A, UBE3A, UBR1, UNC80, UPB1, UPF3B, UQCRB, UQCRCQ, UROC1, USP9X, VLDLR, VPS13B, VPS33B, VRK1, WAC, WDR45, WDR62, WDR73, WDR81, WNT5A, WWOX, XPA, XPNPEP3, XYLT1, ZBTB16, ZBTB18, ZBTB20, ZBTB24, ZC4H2, ZDHHC9, ZEB2, ZFP57, ZFYVE26, ZIC2, ZMIZ1, ZMYM3, ZMYND11, ZNF711</p> |  |  |  |  |  |
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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                      | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
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| <b>Epileptische Enzephalopathie;</b> Gene: AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DCX, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FGF12, FOXG1, FRRS1L, GABRA1, GABRA2, GABRA5, GABRB1, GABRB3, GABRG2, GABRR2, GAD1, GLS, GNAO1, GOT2, GPHN, GRIN2B, GUF1, HCN1, HNRNPU, IQSEC2, ITPA, KCNA2, KCNB1, KCNQ2, KCNT1, KCNT2, MECP2, NECAP1, NEUROD2, NEXMIF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PLCB1, PNKP, PNPO, POLG, PPP3CA, RANGAP1, RHOTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC6A1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 23                                                    |
| <b>Fallot-Tetralogie;</b> Gene: ABL1, ALDH1A2, CRELD1, CRKL, DISP1, FLT4, FOXC2, GATA4, GATA5, GATA6, GDF1, HAND2, ISL1, JAG1, MYH6, MYOM2, NFATC1, NKX2-5, NOTCH1, NOTCH2, NR2F2, PLXND1, SEMA3D, SMAD2, TAB2, TBX1, TBX5, TRAF7, WNT5A, ZFAND5, ZFPM2, ZIC3                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Apr 24                                                    |
| <b>Fiebersyndrome, hereditär;</b> Gene: ADA2, APOA1, APOA2, APOC2, APOC3, CARD14, ELANE, FGA, GSN, IL1RN, IL36RN, LPIN2, LYZ, MEV, MVK, NLR4, NLRP3, NLRP12, NOD2, NTRK1, OSMR, OTULIN, PLCG2, POMP, PSMB8, PSTPIP1, RIPK1, STING1, TNFAIP3, TNFRSF1A, TRNT1, TTR, WDR1                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Apr 24                                                    |
| <b>Frontotemporale Demenz;</b> Gene: CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Aug 25                                                    |
| <b>Genodermatosen;</b> Gene: A2M, AAGAB, ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, ATP2A2, CARD14, CASP14, CAST, CDSN, CERS3, COG6, COL14A1, CTSC, CYP4F22, DSC2, DSG1, DSP, EBP, ENPP1, FGFR1, FLG, GJA1, GJB2, GJB3, GJB4, GJB6, GRHL2, JUP, KANK2, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A, KRT6B, KRT6C, KRT9, LIPN, LORICRIN, MBTPS2, MPLKIP, NIPAL4, NLRP1, NSDHL, PHYH, PKP1, PNPLA1, POMP, PORCN, PTEN, RHBDF2, RSPO1, SDR9C7, SERPINB7, SLC27A4, SLURP1, SNAP29, SPINK5, SRD5A3, ST14, STS, SULT2B1, TGM1, TGM5, TRPV3, USB1, VPS33B, WNT10A                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mai 24                                                    |
| <b>Glaukom, hereditär;</b> Gene: ADAMTS10, ADAMTS17, ASB10, BEST1, CANT1, COL18A1, COL4A1, CPAMD8, CREBBP, CYP1B1, FOXC1, FOXD3, FOXE3, GJA1, GPATCH3, IFIH1, LMX1B, LTBP2, LOXL1, MYOC, NTF4, OCRL, OPTN, PAX6, PITX2, PITX3, RIGI, SBF2, SH3PXD2B, TBK1, TEK, WDR36                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 25                                                    |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                      | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
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| <b>Gliedergürtel-Muskeldystrophie/LGMD</b> ; Gene: <i>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, ETTDH, FHL1, FKRP, FKTN, FLNC, GAA, GBE1, GFPT1, GMPBP, 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</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 22                                                    |
| <b>Hydrozephalus, hereditär</b> ; Gene: <i>AKT3, AP1S2, ARSB, ARX, B3GALNT2, B3GLCT, B3GNT2, B4GAT1, BLTP1, BUB1B, CC2D2A, CCDC88C, CCND2, CENPF, CEP83, CILK1, COL4A1, CRB2, CRPPA, DAG1, DENND5A, DHCR24, EBP, EEF2, EML1, ERF, FAM20C, FANCB, FGFR1, FGFR2, FGFR3, FKRP, FKTN, FLNA, FLVCR2, GFAP, GLI3, GPM2, GUSB, HDAC6, HYL1, IDS, KIAA0586, KIDINS220, KIF7, L1CAM, LAMB1, LARGE1, MAN2B1, MPDZ, MTM1, MYMK, NF1, NSD1, OSTM1, P4HB, PIK3R2, PLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP2R5D, PTCH1, PTCH2, PTEN, RNF125, RPS6KA3, RXYLT1, SEC24D, SKI, SMARCC1, SNX10, STRADA, SUFU, SUMF1, TBC1D32, TCF12, TCIRG1, TNFRSF11A, TRIM71, TWIST1, USP9X, WASHC5, WNT3, ZBTB20, ZIC2, ZIC3</i>                            | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 24                                                    |
| <b>Hypercholesterinämie, hereditär</b> ; Gene: <i>LDLR (NM_000527.5), APOB (NM_000384.3), LDLRAP1 (NM_015627.3), PCSK9 (NM_174936.4)</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mai 23                                                    |
| <b>Hyperkalzämie, hereditär</b> ; Gene: <i>AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mai 24                                                    |
| <b>Hyperkeratosen</b> , hereditär; Gene: <i>AAGAB, AQP5, ATP2A2, ATP2C1, CAST, CDSN, CHST8, CSTA, CTSC, DSG1, DSP, ENPP1, FLG2, GJB2, GJB4, GJB6, JUP, KRT1, KRT14, KRT16, KRT17, KRT6A, KRT6B, KRC6C, KRT9, MBTPS2, PKP1, SERPINB7, SERPINB8, SLURP1, SMARCAD1, TGM5, TRPV4, WNT10A</i>                                                                                                                                                                                                                                                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mai 24                                                    |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                      | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
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| <b>Hyperparathyreoidismus, primär;</b> Gene: <i>CASR, CDC73, CDKN1B, MEN1, RET</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                    | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 21                                                    |
| <b>Hypogonadotroper Hypogonadismus (Kallmann-Syndrom);</b> Gene: <i>ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NROB1, NSMF, PROK2, PROKR2, SEMA3A, SOX10, SPRY4, TAC3, TACR3, TCF12, WDR11</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jul 24                                                    |
| <b>Hypophysenadenom, hereditär;</b> Gene: <i>CDKN1B (NM_004064.5); MEN1 (NM_001370259.2), NF2 (NM_000268.4), PRKAR1A (NM_002734.5)</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Apr 25                                                    |
| <b>Immundefizienz, primär;</b> Gene: <i>ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, ALPI, ALPK1, ANGPT1, AP1S3, AP3B1, AP3D1, ARPC1B, ATAD3A, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFI, CFP, CHD7, CIB1, CIITA, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1B, DCLRE1C, DEF6, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, EFL1, ELANE, ELF4, EPG5, ERBIN, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCGR3A, FCHO1, FERMT1, FERMT3, FNIP1, FOXP1, FOXP3, FPR1, G6PC3, G6PD, GATA1, GATA2, GFI1, GIMAP5, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HPS1, HPS4, HPS6, HTRA2, ICOS, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IPO8, IRAK4, IRF3, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, IVNS1ABP, JAGN1, JAK3, KDM6A, KMT2A, KMT2D, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MBL2, MCM4, MEFV, MOGS, MPEP1, MPO, MRTFA, MSN, MTHFD1, MVK, MYD88, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLR4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NPC1, NRAS, NSMCE3, OAS1, ORAI1, OTULIN, PARN, PAX1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLG, PNP, POLA1, POLD1, POLE, POLR3A, POLR3C, POMP, PRF1, PRIM1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSTPIP1, PTEN, PTPN2, PTPRC, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RC3H1, RECQL4, RELA, RFX5, RFXANK, RFXAP,</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0,<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jul 23                                                    |

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| RHBDF2, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU7-1, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SEC61A1, SERPING1, SGPL1, SH2D1A, SKIC2, SKIC3, SLC27A4, SLC29A3, SLC35C1, SLC39A7, SLC46A1, SLC7A7, SLC9A3, SLCO2A1, SMARCA1, SMARCD2, SNORA31, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT5B, STIM1, STING1, STK4, STX11, STXBP2, STXBP3, SYK, TAFAZZIN, TAP1, TAP2, TAPBP, TBK1, TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TGFB1, TGFB2, TICAM1, TINF2, TLR3, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13C, TNFRSF1A, TNFRSF9, TOP2B, TPP2, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC7A, TYK2, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, XIAP, ZAP70, ZBTB24, ZNF341, ZNFX1                                                                                                                             |                            |                                                                                                                                                     |                                                                                                                                         |                                                   |                                                           |
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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                     | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
| <b>Joubert-Syndrom und Differenzialdiagnosen;</b> Gene: ARL13B, ARL3, ARMC9, ATXN10, B9D1, B9D2, C2CD3, CELSR2, CEP41, CEP55, CPLANE1, CSPP1, EXOC8, HYL51, IFT172, INPP5E, MKS1, NPHP3, PDE6D, PIBF1, POC1B, SUFU, TCTN1, TCTN3, TMEM107, TMEM138, TMEM237                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mai 25                                                    |
| <b>Juvenile Makuladegeneration/M. Stargardt;</b> 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,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 24                                                    |
| <b>Lynch-Syndrom (HNPCC),</b> Gene: MLH1, MSH2, MSH6, PMS2, EPCAM                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) |                                                           |
| <b>Kardiomyopathie (v2);</b> 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, FKR, 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, | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 24                                                    |

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| 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                                                                                                                                                                                                                                                                                                                                            |                            |                                                                                                                                                     |                                                                                                                                            |                                                   |                                                           |
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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                        | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
| <b>Katarakt, hereditär;</b> 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,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1)<br>Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jul 24                                                    |
| <b>Kavernome, hereditär;</b> Gene: CCM2 (NM_031443.4), COL4A1 (NM_001845.6), EPHB4 (NM_004444.5), KRIT1 (NM_194454.3), PDCD10 (NM_007217.4), RASA1 (NM_002890.3)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1)<br>Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mai 23                                                    |
| <b>Kleinwuchs, hereditär;</b> Gene: ACAN, ACP5, AGPS, AKT1, ALPL, ANKH, ANOS, ARID1A, ARID1B, ARSL, ATR, ATRX, B3GALT6, BLM, BMP2, BMP4, BMPR1A, BMPR1B, BTK, CANT1, CCDC8, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CHST14, CHST3, CILK1, COL10A1, COL11A1, COL11A2, COL1A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREBBP, CTSK, CUL7, DDR2, DHCR24, DHCR7, DLL3, DYM, DYNC2H1, EBP, EIF2AK3, EP300, ERCC6, ERCC8, EVC, EVC2, EXT1, EXT2, EYA1, FAM111A, FAM20C, FBN1, FGD1, FGF3, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FOXL2, GDF5, GH1, GHR, GHRH, GHRHR, GHSR, GLI2, GLI3, GPC6, HDAC8, HES7, HESX1, HSPG2, IFT122, IFT140, IFT80, IGF1, IGF1R, IGF2, IGFALS, IHH, INSR, IRS1, KDM6A, KIF22, KMT2D, KRAS, LFNG, LHX3, LHX4, LIFR, MATN3, MESP2, MMP13, MMP9, NBAS, NBN, NEK1, NIPBL, NKX3-2, NOTCH2, NPR2, NR5A1, OBSL1, ORC1, ORC4, ORC6, OTX2, PAPSS2, PCNT, PIK3R1, PITX2, POU1F1, PRKAR1A, PROP1, PTCH1, PTH1R, PTPN11, RAD21, RAF1, RBBP8, RMRP, RNU4ATAC, ROR2, RPS6KA3, RUNX2, SH3PXD2B, SHH, SHOX, SIX3, SLC26A2, SLC35D1, SLC39A13, SMARCA1, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SOS1, SOX2, SOX3, SOX9, SRCAP, STAT5B, SULF1, | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1)<br>Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 23                                                    |

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| TBCE, TBX19, TBX3, TBX6, TGIF1, THRB, TRIM37, TRIP11, TRPS1, TRPV4, WDR35, WNT5A, WRN, ZIC2                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |                            |                                                                                                                                    |                                                                                                                          |                                             |                                                           |
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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                               | Anweisung (Version)<br>Pipeline/Kit                                                                                      | Gerät                                       | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
| <b>Kraniosynostosen, hereditär;</b> 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, HNRNP, 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, TMC01, 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) | Dez 23                                                    |
| <b>Leukodystrophie und Leukoencephalopathie;</b> 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) | Feb 24                                                    |
| <b>Lysosomale Speichererkrankungen,</b> Gene: AGA, ARSA, ARSB, ARSG, ASAH1, ATP13A2, CLCN7, CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSF, CTSK, DNAJC5, FUCA1, GAA, GALC, GALNS, GBA1, GLA, GLB1, GM2A, GNE, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, TPP1, VPS16, VPS33A                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | 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) | Dez 24                                                    |
| <b>Magenkarzinom, diffuses,</b> 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) | Nov 20                                                    |
| <b>Makuladystrophie;</b> 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, RDHS, 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) | Nov 24                                                    |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                     | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------|-----------------------------------------------------------|
| <b>Makrozephalie;</b> Gene: <i>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, SPRED1, STRADA, SUFU, SUZ12, SYN1, TBC1D7, TCF20, TMC01, TMEM94, UPF3B, WNT5A, ZBTB20, ZDHHC9</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 23                                                    |
| <b>Maligne Hyperthermie;</b> Gene: <i>CACNA1S, RYR1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 23                                                    |
| <b>Mamma- und Ovarialkarzinom (HBOC), 12 Gene:</b> <i>ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, PALB2, RAD51C, RAD51D, PTEN, STK11, TP53</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Sep 24                                                    |
| <b>Mamma- und Ovarialkarzinom (HBOC), 6 Gene:</b> <i>BRCA1, BRCA2, CHEK2, PALB2, RAD51C, ATM</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Jan 24                                                    |
| <b>Mamma- und Ovarialkarzinom,</b> Gene: <i>BRCA1, BRCA2</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Okt 20                                                    |
| <b>Marfan-Syndrom und Loeys-Dietz-Syndrom,</b> Gene: <i>FBN1, TGFB1, TGFB2</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Okt 20                                                    |
| <b>Meckel-Gruber-Syndrom und Differenzialdiagnosen;</b> Gene: <i>AHI1, ARL13B, B9D1, B9D2, CC2D2A, CEP120, CEP290, CEP41, CEP55, CPLANE1, CSPP1, CHCR7, KIAA0586, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PIBF1, RPGRIP1L, TCTN1, TMEM107, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B,</i>                                                                                                                                                                                                                                                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-                                           | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:                                      | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Feb 25                                                    |

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| TXNDC15, WDPCP                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |                        | Analyse; PCR, Sanger-Sequenzierung                                                                                                 | varfeed 2.5.1, Limbus, Rostock                                                                                           |                                             |                                                   |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------|------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------|---------------------------------------------|---------------------------------------------------|
| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | Untersuchungs-material | Untersuchungstechnik                                                                                                               | Anweisung (Version) Pipeline/Kit                                                                                         | Gerät                                       | Erstellt/im akkreditierten Bereich aufgenommen am |
| <b>Melanom, hereditär:</b> BAP1, CDK4, CDKN2A, POT1, PTEN, TP53                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | 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) | Feb 23                                            |
| <b>Mikrozephalie, hereditär;</b> 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, CTNBNB1, 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, 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, VP550, VRK1, WDR11, WDR37, WDR4, WDR62, WDR73, XRCC4, YIF1B, YIPF5, ZEB2, ZNF335, ZNF526, ZNF668 | 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) | Okt 23                                            |
| <b>Mitochondriopathie, nukleär;</b> Gene: ABAT, ABCB7, ACAD9, ACO2, AFG3L2, AGK, AIFM1, ANO10, APTX, ATAD3A, ATP5F1A, ATP5F1B, ATP5F1C, ATP5F1D, ATP5F1E, ATP5MC1, ATP5MC2, ATP5MC3, ATP5PO, ATPAF1, ATPAF2, BCS1L, BOLA3, BTD, C1QBP, C2orf69, CA5A, CAR52, CEP89, CHCHD10, CLPB, CLPP, COA1, COA3, COA4, COA5, COA6, COA7, COA8, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX11, COX14, COX15, COX16, COX17, COX18, COX19, COX20, COX4I1, COX4I2, COX5A, COX5B, COX6A1, COX6A2, COX6B1, COX6B2, COX6C, COX7A1, COX7B, COX7C, COX8A, CRLS1, CYC1, DARS2, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNMT1L, DNM2, EARS2, ECHS1, ELAC2, ERAL1, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FDXR, FH, FLAD1, GATB, GATC, GDAP1, GFER, GFM1, GFM2, GLRX5, GTPBP3, HARS2, HCCS, HIBCH, HLCS, HPDL, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH3A, IDH3B, ISCA1, ISCA2, ISCU, KARS1, LARS2, LIAS, LIG3, LIPT1, LIPT2, LONP1, LRPPRC,                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | 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) | Jul 23                                            |

## Liste

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |                               |                                                                                                                                    |                                                                                                                          |                                             |                                                          |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------|------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------|---------------------------------------------|----------------------------------------------------------|
| LYRM4, LYRM7, MARS2, MDH2, MECR, MFF, MFN2, MGME1, MICOS13, MICU1, MIPEP, MPC1, MPV17, MRM2, MRPL12, MRPL3, MRPS14, MRPS16, MRPS23, MRPS34, MRPS7, MSTO1, MTFMT, MTO1, MTPAP, MTRFR, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA3, NDUFA4, NDUFA5, NDUFA6, NDUFA7, NDUFA8, NDUFA9, NDUFAB1, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFAF8, NDUFB1, NDUFB10, NDUFB11, NDUFB2, NDUFB3, NDUFB4, NDUFB5, NDUFB6, NDUFB7, NDUFB8, NDUFB9, NDUFC1, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFS1, NFU1, NSUN3, NUBPL, OPA1, OPA3, OXA1L, PARS2, PC, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PET117, PMPCA, PMPCB, PNPLA8, PNPT1, POLG, POLG2, POLRMT, PPA2, PTCO3, PUS1, QRS1, RARS2, RMND1, RNASEH1, RRM2B, RTN4IP1, SACS, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHAF3, SDHAF4, SDHB, SDHC, SDHD, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC25A1, SLC25A12, SLC25A19, SLC25A21, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SPG7, SQOR, SSBP1, SUCLA2, SUCLG1, SURF1, TACO1, TAFAZZIN, TARS2, TFAM, TIMM22, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126B, TMEM65, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCC2, UQCC3, UQCRB, UQCRC1, UQCRC2, UQCRCF1, UQCRH, UQCRCQ, VARS2, YARS2, YME1L1 |                               |                                                                                                                                    |                                                                                                                          |                                             |                                                          |
| <b>Analyt (Messgröße)</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | <b>Untersuchungs-material</b> | <b>Untersuchungstechnik</b>                                                                                                        | <b>Anweisung (Version) Pipeline/Kit</b>                                                                                  | <b>Gerät</b>                                | <b>Erstellt/im akkreditierten Bereich aufgenommen am</b> |
| <b>MODY</b> , 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) | Mrz 21                                                   |
| <b>Motoneuropathie</b> ; Gene: AAAS, AARS1, ABCA1, ABHD12, ACTA1, ADAR, AGRN, AIFM1, ALG14, ALG2, ALS2, AMACR, ANG, ANOS, ANXA11, ARHGEF10, ASAH1, ASCC1, ATL1, ATL3, ATM, ATP1A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSLC12, CACNA1A, CADM3, CAPN3, CASQ1, CAVIN1, CCDC78, CCT5, CD59, CFL2, CHAT, CHCHD10, CHKB, CHMP2B, CHRNA1, CHRN1, CHRN2, CHRN3, CLCF1, CLP1, CNTN1, CNTNAP1, COA7, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX10, COX6A1, CRLF1, CRPPA, CTDP1, CYP27A1, DAG1, DCAF8, DCTN1, DCTN2, DES, DGAT2, DHTKD1, DMD, DNAJB2, DNAJB5, DNMT2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DRP2, DST, DYNC1H1, DYSF, EGR2, ELP1, EMD, EPG5, ERLIN1, EXOSC3, EXOSC8, FBLN5, FBXO38, FGD4, FHL1, FIG4, FKBP14, FKRP, FKTN, FUS, FXN, GAA, GALT, GAN, GARS1, GBE1, GBF1, GDAP1, GFPT1, GJB1, GLA, GLDC, GMPPB, GNB4, GSN, GYG1, HADHA, HADHB, HARS1, HEXA, HINT1, HK1, HMBS, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB1, HSPB3, HSPB8, HYCC1, IFRD1, IGHMBP2, INF2, ITGA7, ITPR3, JAG1, JPH1, KARS1, KBTBD13, KIF1A, KIF1B, KIF5A, KLHL13, KLHL40, KY, LAMA2, LAMB2, LAMP2, LARGE1, LAS1L, LIMS2, LITAF, LMNA, LMOD3, LRP4, LRSAM1, MARS1, MATR3, MCM3AP, MED25, MEGF10, MFN2, MICU1, MME, MORC2, MPV17, MPZ, MTM1, MTMR2, MTRFR, MUSK, MYH14, MYH7                                                                                                         | 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) | Feb 25                                                   |

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| MYO18B, MYO9A, NAGLU, NDRG1, NDUFAF5, NEB, NEFH, NEFL, NEK1, NGF, NIPA1, NTRK1, OPTN, ORAI1, PDHA1, PDK3, PEX12, PFN1, PIEZO2, PLEC, PLEKHG5, PLP1, PMP22, PNKP, POLG, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRDM12, PREPL, PRPS1, PRX, PTRH2, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RXYLT1, RYR1, SACS, SARS1, SBDS, SBF1, SBF2, SCN10A, SCN11A, SCN4A, SCN9A, SCO2, SELENON, SEPTIN9, SETX, SGCA, SGCB, SGCD, SGCG, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC16A2, SLC25A1, SLC25A21, SLC25A26, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMAD3, SMN1, SNAP25, SOD1, SORD, SPAST, SPG11, SPTLC1, SPTLC2, SPTLC3, SQSTM1, STIM1, SURF1, SYNE1, SYT2, TARDBP, TBK1, TCAP, TDP1, TECPR2, TFG, TIA1, TK2, TNNT1, TNNT2, TNNT3, TNPO3, TNXB, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRIP4, TRPA1, TRPV4, TTN, TTR, TUBA4A, TUBB3, TYMP, UBA1, UBQLN2, UNC13A, VAPB, VCP, VMA21, VRK1, VWA1, WNK1, YARS1 |                            |                                                                                                                                                      |                                                                                                                                         |                                                   |                                                           |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------|-----------------------------------------------------------|
| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                 | Anweisung (Version)<br>Pipeline/Kit                                                                                                     | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
| <b>Mukopolysaccharidosen;</b> Gene: AGA, ARSB, CTSA, FUCA1, GALNS, GBA1, GLA, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MAN2B1, MANBA, MCOLN1, NAGA, NAGLU, NEU1, PSAP, SGSH, SLC17A5                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung  | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 23                                                    |
| <b>Multiple Endokrine Neoplasie Typ 1 /4,</b> Gene: CDC73, CDKN1B, MEN1                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis);<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Aug 20                                                    |
| <b>Multiple Endokrine Neoplasie Typ 2,</b> Gene: RET                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung  | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Aug 20                                                    |
| <b>Multiple epiphysäre Dysplasie und Pseudoachondroplasie;</b> Gene: COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung  | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 23                                                    |
| <b>Myopathien, hereditär;</b> 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, CHRN1, CHRN1                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-                                            | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:                                      | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 23                                                    |

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| CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL13A1, COL25A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A2, CPT1A, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DHX16, DMD, DNA2, DNAJB4, DNAJB6, DNMT2, 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, HMGR, 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, 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 |                               | Analyse; PCR, Sanger-Sequenzierung                                                                                                 | varfeed 2.5.1, Limbus, Rostock                                                                           |                                             |                                                          |
| <b>Analyt (Messgröße)</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | <b>Untersuchungs-material</b> | <b>Untersuchungstechnik</b>                                                                                                        | <b>Anweisung (Version) Pipeline/Kit</b>                                                                  | <b>Gerät</b>                                | <b>Erstellt/im akkreditierten Bereich aufgenommen am</b> |
| <b>Neuroendokrine Tumore / NET;</b> Gene: <i>CDKN1B, MEN1, NF1, POT1, RET, TSC1, TSC2, VHL</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | 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) | Nov 24                                                   |
| <b>Neurofibromatose Typ 1 / M. Recklinghausen:</b> Gen: <i>NF1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | 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) | Dez 20                                                   |
| <b>Neurofibromatose Typ 2:</b> Gen: <i>NF2</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | 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) | Dez 20                                                   |
| <b>Neuropathie, hereditär;</b> Gene: <i>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,</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | EDTA-Blut, DNA                | Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline:                                                               | AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0                                        | Illumina NextSeq, SeqStudio (Thermo Fisher) | Aug 23                                                   |

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|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------|------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------|---------------------------------------------|----------------------------------------------------------|
| CD59, CFAP276, CHCHD10, CNTNAP1, COA7, COX6A1, COX10, CPOX, CTDPI, 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, GALT, 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, MTPP, 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, 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, ZFX2, ZFYVE26 |                               | varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung                                                                      | Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock                                                                   |                                             |                                                          |
| <b>Analyt (Messgröße)</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | <b>Untersuchungs-material</b> | <b>Untersuchungstechnik</b>                                                                                                        | <b>Anweisung (Version) Pipeline/Kit</b>                                                                                  | <b>Gerät</b>                                | <b>Erstellt/im akkreditierten Bereich aufgenommen am</b> |
| <b>Neutropenie, hereditär;</b> Gene: ADA2, CD40, CD40LG, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GF11, GINS1, HAX1, IFNGR2, JAGN1, LAMTOR2, LYST, PGM3, RAC2, SBDS, SMARCD2, SRP54, SRP72, TAFAZZIN, TCIRG1, TLR8, USB1, VPS45, WAS, WDR1, WIPF1                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | 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) | Sep 23                                                   |
| <b>Nierenerkrankungen, hereditär;</b> Gene: ACE, ACTN4, AGT, AGTR1, AGXT, ALMS1, ANLN, ANOS1, APOA1, APOL1, APRT, ARHGAP24, ARHGDIA, ATP6V0A4, ATP6V1B1, ATP7B, BICC1, BMP4, BMP7, BSND, CA2, CACNA1D, CACNA1H, CASR, CD151, CD2AP, CDC5L, CDC73, CHD1L, CHRM3, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN14, CLDN16, CLDN19, COA5, COA8, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, COX10, COX6B1, CRB2, CRKL, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, DGKE, DMP1, DSTYK, EHHADH, EMP2, ENPP1, EYA1, FAM20A, FASTKD2, FAT1, FGA, FGF20, FGF23, FN1, FOXI1, FRAS1, FREM1, FREM2, G6PC1, GATA3, GCM2, GDNF, GLA, GLIS3, GREM1, GRHPR, GRIP1, GTF2I, GUCY2C, HNF1B, HNF4A, HOGA1, HPRT1, HPSE2, HRAS, HSD11B2, INF2, INSR, ITGA3, ITGA8, JAG1, KANK1, KANK2, KANK4, KAT6B, KCNJ1, KCNJ5, KLHL3, LAMB2, LIFR, LMX1B, LRIG2, LRP2, LRP4, LYZ, MAGED2, MEFV, MEN1, MMACHC, MOCOS, MUC1, MYH9, MYO1E, NHERF1, NOTCH2, NPHS1, NPHS2, NR3C1, NR3C2, NRIP1, NUP107, NUP205, NUP93, NXF5, OCRL, PAX2, PBX1, PDE3A, PDSS2, PHEX, PLCE1, PMM2, PPARG, PRPS1, PTH1R, PTPRO, RAD51C, REN, RET, RMND1, ROBO2, SALL1, SALL4, SBDS, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC61A1, SGPL1, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC26A1, SLC2A2,                                                                                                                        | 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) | Mrz 21                                                   |

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| SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC3A1, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9, SMARCA1, SOX17, STRA6, TACO1, TBC1D1, TBC1D24, TBX18, TMEM127, TNXB, TRAP1, TRPC6, UMOD, UPK3A, VPS33B, WNK1, WNK4, WNT4, WT1, XDH, XPO5                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |                            |                                                                                                                                                     |                                                                                                                                         |                                                   |                                                           |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------|-----------------------------------------------------------|
| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                | Anweisung (Version)<br>Pipeline/Kit                                                                                                     | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
| <b>Nierenzellkarzinom, hereditär</b> , Gene: <i>BAP1, FLCN, FH, MET, PTEN, SDHB, SDHD, TSC1, TSC2, VHL</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Nierenzellkarzinom, papilläres</b> , Gen: <i>MET</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Noonan-Syndrom</b> , Gene: <i>PTPN11, BRAF, KRAS, RAF1, RIT1, SOS1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 20                                                    |
| <b>Nystagmus, hereditär</b> ; Gene: <i>AHR, AP1B1, AP3B1, BLOC1S3, BLOC1S5, BLOC1S6, CACNA1A, CACNA1F, CASK, EP290, CNGA3, CNGB3, DAGLA, DTNBP1, FRMD7, GPR143, GUCY2D, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, MANBA, MLPH, NYX, OCA2, PAX6, RDH12, RPE65, SACS, SETX, SLC24A5, SLC38A8, SLC45A2, TULP1, TYR, TYRP1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jan 24                                                    |
| <b>Okuläre Fehlbildungen und Optikushypoplasie</b> ; Geme: <i>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, PDS51, 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</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Jun 23                                                    |
| <b>Okulärer und Okulokutater Albinismus</b> ; Gene: <i>DCT, GPR143, LRMDA, OCA2, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-                                           | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:                                      | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 25                                                    |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | Untersuchungs-<br>material | Analyse; PCR, Sanger-<br>Sequenzierung<br><br><b>Untersuchungstechnik</b>                                                                           | varfeed 2.5.1, Limbus,<br>Rostock<br><br><b>Anweisung (Version)<br/>Pipeline/Kit</b>                                                    | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
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| <b>Optikusatrophie, hereditäre</b> ; Gene: <i>ABCA4, ACO2, AFG3L2, ALDH1A3, ALPK1, ANTXR1, AP3B2, ATG7, ATP1A3, AUH, B3GALNT2, BLOC1S1, C19orf12, CISD2, COL4A1, COL4A2, DDHD2, DNAJC19, DNAJC30, DNM1L, ELOVL4, EPRS1, FA2H, FDXR, HESX1, HIKESHI, HK1, ISCA2, KANSL1, KIF7, KNSTRN, MAG, MCAT, MECR, MFF, MFN2, MIEF1, MTPAP, MTRFR, NARS2, NBAS, NDUFA12, NR2F1, OPA1, OPA3, OTX2, PAX2, PAX6, PDSS1, PIK3CD, POLG, PROM1, PRPH2, PRPS1, PUF60, RTN4IP1, SLC19A2, SLC25A46, SLC38A8, SLC44A1, SLC52A2, SNX10, SOX2, SOX5, SPG7, SSBP1, TFG, TIMM8A, TMEM126A, TSFM, TUBA8, UBE3B, UCHL1, VAX1, WFS1, YME1L1</i> | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Feb 24                                                    |
| <b>Osteogenesis imperfecta</b> ; Gene: <i>ALPL; B3GALT6; B4GALT7; BMP1; COL10A1; COL11A1; COL11A2; COL1A1; COL1A2; COL2A1; COPB2; CREB3L1; CRTAP; DMP1; DSPP; FGF23; FKBP10; GORAB; IFITM5; KDELR2; MBTPS2; MESD; NOTCH2; P3H1; P4HB; PHEX; PLOD2; PLS3; PPIB; PYCR1; SEC24D; SERPINF1; SERPINH1; SGMS2; SLC34A3; SP7; SPARC; SUCO; TAPT1; TENT5A; TMEM38B; TNFRSF11A; TRPV6; UNC45A; WNT1; WNT11</i>                                                                                                                                                                                                              | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Sep 23                                                    |
| <b>Osteopetrose</b> ; Gene: <i>AMER1, ANKH, CA2, CLCN7, CTSK, FAM20C, FERMT3, HPGD, LEMD3, LRP5, MTAP, OSTM1, PLEKHM1, SNX10, SOST, TCIRG1, TNFSF11, TYROBP</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                    | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Okt 24                                                    |
| <b>Oslerische Krankheit / Morbus Osler / Hereditäre hämorrhagische Teleangiektasie (HHT)</b> ; Gene: <i>ACVRL1 (NM_000020.3), BMPR2 (NM_001204.7), ENG (NM_00114753.3), EPHB4 (NM_004444.5), GDF2 (NM_016204.4), RASA1 (NM_002890.3), SMAD4 (NM_005359.6), SOX18 (NM_018419.3)</i>                                                                                                                                                                                                                                                                                                                                 | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 23                                                    |
| <b>Pankreatitis, hereditär</b> : <i>PRSS1, SPINK1, CPA1, CTSC, CASR</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 20                                                    |
| <b>Pankreaskarzinom</b> , Gene: <i>ATM, BRCA1, BRCA2, CDKN2A, MLH1, MSH6, PALB2, STK11, TP53</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 20                                                    |
| <b>Paragangliom-Phäochromozytom-Syndrom</b> , Gene: <i>AIP, MAX, MEN1, NF1, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, TMEM127, VHL</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:                                                                          | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-                                                                  | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Nov 20                                                    |

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| <b>Analyt (Messgröße)</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | <b>Untersuchungs-material</b> | <b>Untersuchungstechnik</b>                                                                                                        | <b>Anweisung (Version) Pipeline/Kit</b>                                                                                  | <b>Gerät</b>                                | <b>Erstellt/im akkreditierten Bereich aufgenommen am</b> |
| <b>Paraplegie / Hereditäre Spastische Paraplegie (HSP);</b> Gene: <i>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, DNMT2, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FBXO7, FIG4, GAD1, GALT, 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, TNFR, TUBB4A, UBAP1, UBQLN2, UCHL1, VAMP1, VAPB, VCP, VPS37A, WASHC5, WDR45B, WDR48, ZFYVE26, ZFYVE27</i> | 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) | Sep 24                                                   |
| <b>Parkinson-Syndrom, hereditär;</b> Gene: <i>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</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       | 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) | Mai 23                                                   |
| <b>Peutz-Jeghers-Syndrom,</b> Gen: <i>STK11</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | 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) | Dez 20                                                   |
| <b>Polyposis coli,</b> Gene: <i>APC, BMPR1A, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | 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) | Nov 20                                                   |
| <b>Porphyrie, hereditär;</b> Gene: <i>ABCC2, ALAD, ALAS2, CDIN1, CLPX, CPOX, FECH, GATA1, HFE, HMBS, PPOX, SLC19A2, UROD, UROS</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | 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) | Sep 24                                                   |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                                   | Anweisung (Version)<br>Pipeline/Kit                                                                                                     | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
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| <b>Prostatakarzinom:</b> <i>ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH6, PMS2, RAD51C, TP53</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Jun 22                                                    |
| <b>RASopathie,</b> Gene: <i>A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1</i>                                                                                                                                                                                                                                                                                                                                                                                                                   | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Rhabdoid-Prädispositions-Syndrom,</b> Gene: <i>SMARCB1, SMARCA4</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Schilddrüsenkarzinom, medulläres,</b> Gen: <i>RET</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 21                                                    |
| <b>Schilddrüsenkarzinom, papilläres,</b> Gene: <i>APC, CDKN1B, DICER1, PRKAR1A, PTEN</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Feb 23                                                    |
| <b>Schwannomatose,</b> Gene: <i>LZTR1, NF2, SMARCB1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                   | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Schwerhörigkeit, hereditär;</b> Gene: <i>ABCC1, ABHD12, ACTG1, ADGRV1, AFG2A, AFG2B, AIFM1, ANKH, ARSG, ATOH1, ATP1A3, ATP11A, ATP2B2, ATP6V1B1, BDP1, BSND, BTB, 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, FGF3, FITM2, FOXI1, GAB1,</i> | EDTA-Blut, DNA             | Capture Enrichment (Twist<br>Exome 2.0), Sequencing-by-<br>synthesis, Bioinformatik-<br>Pipeline: varfeed; SNV-, Indel-,<br>CNV-Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Sep 24                                                    |

Liste

| 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, TMT2C, TNC, TOP2B, TPRN, TRIOBP, TRRAP, TSPEAR, TWNK, USH1C, USH1G, USH2A, USP48, WBP2, WFS1, WHRN                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |                                    |                                                                                                                                                      |                                                                                                                          |                                             |                                                                    |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------|---------------------------------------------|--------------------------------------------------------------------|
| <b>Analyt (Messgröße)</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | <b>Untersuchungs-<br/>material</b> | <b>Untersuchungstechnik</b>                                                                                                                          | <b>Anweisung (Version)<br/>Pipeline/Kit</b>                                                                              | <b>Gerät</b>                                | <b>Erstellt/im akkre-<br/>ditierten Bereich<br/>aufgenommen am</b> |
| <b>Septo-optische Dyplasie;</b> 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) | Jul 25                                                             |
| <b>Skeletterkrankungen, hereditär;</b> Gene: ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTS12, AFF3, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARID1A, ARID1B, ARID2, ARL6, ARSB, ARSL, ASXL1, ASXL2, ATP6V0A2, ATP7A, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDH3, CDKN1C, CDT1, CEP120, CEP290, CFAP410, CHST14, CHST3, CHSY1, CILK1, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRTAP, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DHCR24, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNAJC21, DNMT3A, DOCK6, DONSON, DPAGT1, DPM1, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EED, EFL1, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXT3, EZH2, FAM111A, FAM20C, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GJA1, GLB1, GLI1, GLI2, GLI3, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GSC, GUSB, GZF1, HDAC8, HES7, HGSNAT, HHAT, HOXA13, HOXD13, HPGD, HS2ST1, HSPA9, HSPG2, IARS2, IDH1, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IHH, IKBKG, IL11RA, IL1RN, INPPL1, IQCE, KAT6B, KDELR2, KIAA0586, KIAA0753, KIAA0825, KIF22, KIF7, KMT2D, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP3, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MBTPS1, MECOM, MEGF8, | 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) | Nov 23                                                             |

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|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------|---------------|
| <p>MEOX1, MESD, MESP2, MGP, MKKS, MKS1, MMP13, MMP2, MMP9, MNX1, MPDU1, MSX2, MTX2, MYCN, MYH3, MYO18B, NAA10, NAGLU, NANS, NBAS, NEK1, NEPRO, NEU1, NFIX, NIPBL, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NSD1, NSDHL, NXN, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRKG2, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RASGRP2, RBM8A, RBPJ, RECQL4, RFT1, RINT1, RMRP, RNU4ATAC, ROR2, RPRIP1L, RPL13, RSPRY1, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCUBE3, SEC24D, SERPINF1, SERPINH1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCA1, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, SRP54, STT3A, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFB2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, UFSP2, UNC45A, VDR, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, YY1, YY1AP1, ZMPSTE24, ZNF687, ZSWIM6</p> |                       |                                                                                                                                                             |                                                                                                                                 |                                                    |               |
| <p><b>Spastische Paraplegie/SPG, hereditär;</b> 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, DNMT2, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FBXO7, FIG4, GAD1, GALT, 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, TNF, TUBB4A, UBAP1, UBQLN2, UCHL1, VAMP1, VAPB, VCP, VPS37A, WASHC5, WDR45B, WDR48, ZFYVE26, ZFYVE27</p>                                                                                                                                                                                                                                                               | <p>EDTA-Blut, DNA</p> | <p>Capture Enrichment (Twist Exome 2.0), Sequencing-by-synthesis, Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung</p> | <p>AA_NGS-NextSeq (v3), AA_Sanger-Sequenzierung (v1) Twist Exome 2.0 Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock</p> | <p>Illumina NextSeq, SeqStudio (Thermo Fisher)</p> | <p>Jun 23</p> |
| <p><b>Thorakale Aortenerweiterung,</b> Gene: ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB2, TGFB2, TGFB2</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | <p>EDTA-Blut, DNA</p> | <p>Capture Enrichment, Sequencing-by-synthesis; Bioinformatik-Pipeline: varfeed; SNV-, Indel-, CNV-Analyse; PCR, Sanger-Sequenzierung</p>                   | <p>AA_NGS_MiniSeq (v5), AA_Sanger-Sequenzierung (v1) Bioinformatik-Pipeline: varfeed 2.5.1, Limbus, Rostock</p>                 | <p>Illumina MiniSeq, SeqStudio (Thermo Fisher)</p> | <p>Jan 24</p> |

Liste

| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Untersuchungs-<br>material | Untersuchungstechnik                                                                                                                                                   | Anweisung (Version)<br>Pipeline/Kit                                                                                                        | Gerät                                             | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------|-----------------------------------------------------------|
| <b>Tuberöse Sklerose</b> , Gene: <i>TSC1</i> , <i>TSC2</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                      | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Usher-Syndrom</b> ; Gene: <i>ABHD12</i> (NM_015600.5), <i>ADGRV1</i> (NM_032119.4), <i>ARSG</i> (NM_014960.5), <i>CDH23</i> (NM_022124.6), <i>CEP250</i> (NM_007186.6), <i>CEP78</i> (NM_001098802.3), <i>CIB2</i> (NM_006383.4), <i>CLRN1</i> (NM_001195794.1), <i>ESPN</i> (NM_031475.3), <i>MYO7A</i> (NM_000260.4), <i>PCDH15</i> (NM_001142763.2), <i>PDZD7</i> (NM_001195263.2), <i>PEX1</i> (NM_000466.3), <i>PEX6</i> (NM_000287.4), <i>TUBB4B</i> (NM_006088.6), <i>USH1C</i> (NM_153676.4), <i>USH1G</i> (NM_173477.5), <i>USH2A</i> (NM_206933.4), <i>WHRN</i> (NM_015404.4)                                                                                                                                                  | EDTA-Blut, DNA             | Capture Enrichment (Twist<br>Exome 2.0), Sequencing-by-<br>synthesis, Bioinformatik-<br>Pipeline: varfeed; SNV-, Indel-,<br>CNV-Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock    | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Sep 23                                                    |
| <b>Von-Hippel-Lindau-Syndrom</b> , Gen: <i>VHL</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | EDTA-Blut, DNA             | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                      | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Whole Exome-Sequenzierung (WES)</b>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | EDTA-Blut, DNA             | Capture Enrichment (Twist<br>Exome 2.0), Sequencing-by-<br>synthesis, Bioinformatik-<br>Pipeline: varfeed; SNV-, Indel-,<br>CNV-Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock    | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Feb 23                                                    |
| <b>Wilms-Tumor</b> : <i>WT1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | EDTA-Blut                  | Capture Enrichment,<br>Sequencing-by-synthesis;<br>Bioinformatik-Pipeline:<br>varfeed; SNV-, Indel-, CNV-<br>Analyse; PCR, Sanger-<br>Sequenzierung                    | AA_NGS_MiniSeq (v5),<br>AA_Sanger-Sequenzierung<br>(v1) Bioinformatik-<br>Pipeline: varfeed 2.5.1,<br>Limbus, Rostock                      | Illumina MiniSeq,<br>SeqStudio<br>(Thermo Fisher) | Dez 20                                                    |
| <b>Xeroderma pigmentosum</b> ; Gene: <i>DDB2</i> , <i>ERCC1</i> , <i>ERCC2</i> , <i>ERCC3</i> , <i>ERCC4</i> , <i>ERCC5</i> , <i>ERCC6</i> , <i>ERCC8</i> , <i>POLH</i> , <i>XPA</i> , <i>XPC</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | EDTA-Blut, DNA             | Capture Enrichment (Twist<br>Exome 2.0), Sequencing-by-<br>synthesis, Bioinformatik-<br>Pipeline: varfeed; SNV-, Indel-,<br>CNV-Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1) Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock    | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Mrz 24                                                    |
| <b>Zapfen- und Stäbchen-Dystrophie</b> , Gene: <i>ABCA4</i> , <i>ADAM9</i> , <i>AIPL1</i> , <i>ALMS1</i> , <i>ATF6</i> , <i>BEST1</i> , <i>CABP4</i> , <i>CACNA1F</i> , <i>CACNA2D4</i> , <i>CDHR1</i> , <i>CEP78</i> , <i>CERKL</i> , <i>CFAP410</i> , <i>CFAP418</i> , <i>CNGA3</i> , <i>CNGB3</i> , <i>CNNM4</i> , <i>CRB1</i> , <i>CRX</i> , <i>GNAT2</i> , <i>GUCA1A</i> , <i>GUCY2D</i> , <i>KCNV2</i> , <i>NMNAT1</i> , <i>PCARE</i> , <i>PCYT1A</i> , <i>PDE6C</i> , <i>PDE6H</i> , <i>PITPNM3</i> , <i>POC1B</i> , <i>PROM1</i> , <i>PRPH2</i> , <i>RAB28</i> , <i>RAX2</i> , <i>RDH12</i> , <i>RDH5</i> , <i>RGS9</i> , <i>RGS9BP</i> , <i>RIMS1</i> , <i>RPGR</i> , <i>RPGRIP1</i> , <i>SEMA4A</i> , <i>TTL5</i> , <i>UNC119</i> | EDTA-Blut, DNA             | Capture Enrichment (Twist<br>Exome 2.0), Sequencing-by-<br>synthesis, Bioinformatik-<br>Pipeline: varfeed; SNV-, Indel-,<br>CNV-Analyse; PCR, Sanger-<br>Sequenzierung | AA_NGS-NextSeq (v3),<br>AA_Sanger-Sequenzierung<br>(v1)<br>Twist Exome 2.0<br>Bioinformatik-Pipeline:<br>varfeed 2.5.1, Limbus,<br>Rostock | Illumina NextSeq,<br>SeqStudio<br>(Thermo Fisher) | Apr 24                                                    |
| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Untersuchungs-             | Untersuchungstechnik                                                                                                                                                   | Anweisung (Version)                                                                                                                        | Gerät                                             | Erstellt/im akkre-                                        |

## Liste

[illegible]

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| Analyt (Messgröße)                                                                                                        | Untersuchungs-<br>material | Untersuchungstechnik | Anweisung (Version)<br>Pipeline/Kit | Gerät                         | Erstellt/im akkre-<br>ditierten Bereich<br>aufgenommen am |
|---------------------------------------------------------------------------------------------------------------------------|----------------------------|----------------------|-------------------------------------|-------------------------------|-----------------------------------------------------------|
| ABCC6-Defizienz, Gen: ABCC6                                                                                               | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Apr 24                                                    |
| Caveolin-Defizienz, Gen: CAV3                                                                                             | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Apr 24                                                    |
| CMT1, Gen: PMP22                                                                                                          | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Dez 24                                                    |
| Fumarat-Hydratase-Defizienz, Gen: FH                                                                                      | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Mai 23                                                    |
| HBOC / Mamma- und Ovarialkarzinom, Gene: <i>BRCA1</i> , <i>BRCA2</i>                                                      | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Okt 20                                                    |
| HBOC / Mamma- und Ovarialkarzinom (HBOC), Gene: <i>BRCA1</i> , <i>BRCA2</i> , <i>CHEK2</i> , <i>PALB2</i> , <i>RAD51C</i> | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Jul 23                                                    |
| Hypercholesterinämie, hereditär; Gen: LDLR                                                                                | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo F isher) | Dez 23                                                    |
| Juvenile Polyposis / JPS; Gene: <i>BMPR1A</i> , <i>PTEN</i> , <i>SMAD4</i>                                                | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Aug 23                                                    |
| Langes QT-Syndrom; Gene: <i>KCNQ1</i> , <i>KCNH2</i> , <i>KCNE1</i> , <i>KCNE2</i> , <i>KCNJ2</i>                         | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Apr 24                                                    |
| Lynch-Syndrom, Gene: <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>PMS2</i> , <i>EPCAM</i>                                 | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Sep 21                                                    |
| Multiple Endokrine Neoplasie Typ 1, Gen: <i>MEN1</i>                                                                      | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Dez 21                                                    |
| Polyposis coli, Gene: <i>APC</i> , <i>MUTYH</i>                                                                           | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Feb 22                                                    |
| Porphyrie; Gene: <i>ALAD</i> , <i>CPOX</i> , <i>FECH</i> , <i>HMBS</i> , <i>PPOX</i> , <i>UROS</i> , <i>UROD</i>          | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Nov 24                                                    |
| Peutz-Jeghers-Syndrom, Gen: <i>STK11</i>                                                                                  | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Jul 21                                                    |
| SHOX-Ausfall, Gen: <i>SHOX</i>                                                                                            | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Apr 24                                                    |
| Tuberöse Sklerose, Gene: <i>TSC1</i> , <i>TSC2</i>                                                                        | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Okt 21                                                    |
| Marfan-Syndrom und Loeys-Dietz-Syndrom, Gene: <i>FBN1</i> , <i>TGFBR1</i> , <i>TGFBR2</i>                                 | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Jan 23                                                    |
| Neurofibromatose Typ 1, Gen: <i>NF1</i>                                                                                   | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Okt 20                                                    |
| Neurofibromatose Typ 1, Gen: <i>NF2</i>                                                                                   | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Okt 20                                                    |
| Ehlers-Danlos-Syndrom, vaskulärer Typ, Gen: <i>COL3A1</i>                                                                 | EDTA-Blut, DNA             | MLPA                 | AA_MLPA (v2)                        | SeqStudio<br>(Thermo Fisher)  | Dez 20                                                    |