



Die Liste der Untersuchungsverfahren ist für beide Konformitätsbewertungsstellen (KBS) gültig

Untersuchungsart:  
Chromosomenanalyse<sup>[Flex C]</sup>

| Analyt (Messgröße)          | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik         | Anweisung+Version<br>Pipeline/Kit/Panel+Version | Gerät                                         | CE-<br>Verfahren | in Haus-<br>Verfahren | Erläuterung zu<br>weiterer<br>Bearbeitung,<br>sofern<br>zutreffend | Datum der<br>Aufnahme/<br>Änderung |
|-----------------------------|-------------------------------------------------------------------|------------------------------|-------------------------------------------------|-----------------------------------------------|------------------|-----------------------|--------------------------------------------------------------------|------------------------------------|
| Angeborener Chromosomensatz | peripheres Blut                                                   | Chromosomenbänderungsanalyse | AA-201, V005; AA-206,<br>V004                   | Zeiss, Axio Imager,<br>Metasystems,<br>Ikaros |                  | x                     |                                                                    | 22.01.2013                         |

Untersuchungsart:  
Molekularbiologische Untersuchungen (Amplifikationsverfahren)<sup>[Flex C]</sup>

| Analyt (Messgröße)        | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik  | Anweisung+Version<br>Pipeline/Kit/Panel+Version                                                               | Gerät                                                                                               | CE-<br>Verfahren | in Haus-<br>Verfahren | Erläuterung zu<br>weiterer<br>Bearbeitung,<br>sofern<br>zutreffend | Datum der<br>Aufnahme/<br>Änderung |
|---------------------------|-------------------------------------------------------------------|-----------------------|---------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------|------------------|-----------------------|--------------------------------------------------------------------|------------------------------------|
| Fragiles X-Syndrom (FMR1) | Blut, DNA; DNA                                                    | Fragmentlängenanalyse | AA-103, V004; AA-156,<br>V004; AA-157, V003; AA-<br>209, V003; AA-248,<br>V003; AA-249, V003;<br>AA-250, V004 | ThermoFisher<br>Scientific,<br>SeqStudio;<br>ThermoFisher<br>Scientific, Genetic<br>Analyzer 3500xl |                  | x                     |                                                                    | 04.05.2024                         |

|                                                                          |                |                       |                                                                      |                                                                                      |   |  |            |
|--------------------------------------------------------------------------|----------------|-----------------------|----------------------------------------------------------------------|--------------------------------------------------------------------------------------|---|--|------------|
| Spinocerebellare Ataxie (SCA) (ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7, TBP) | Blut, DNA; DNA | Fragmentlängenanalyse | AA-103, V004; AA-156, V004; AA-157, V003; AA-209, V003; AA-252, V003 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl | x |  | 13.12.2022 |
| Chorea Huntington (HD) (HTT)                                             | Blut, DNA; DNA | Fragmentlängenanalyse | AA-103, V004; AA-156, V004; AA-157, V003; AA-209, V003; AA-254, V003 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl | x |  | 13.12.2022 |
| Friedreichsche Ataxie (FRDA) (FXN)                                       | Blut, DNA; DNA | Fragmentlängenanalyse | AA-103, V004; AA-156, V004; AA-157, V003; AA-209, V003; AA-253, V003 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl | x |  | 10.02.2023 |
| Prader-Willi-Syndrom/Angelman-Syndrom (UBE3A)                            | Blut, DNA; DNA | MLPA                  | AA-113, V005; AA-153, V005; AA-156, V004; AA-157, V003; AA-209, V003 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl | x |  | 29.08.2018 |
| Beckwith-Wiedemann-Syndrom / Silver-Russell-Syndrom (ICR1, ICR2)         | Blut, DNA; DNA | MLPA                  | AA-113, V005; AA-153, V005; AA-156, V004; AA-157, V003; AA-209, V003 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl | x |  | 20.11.2018 |

|                                                                                                                                                                                                                                                                                                                                      |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Lynch-Syndrom / Hereditäres Nicht-polypooses Kolorektalkarzinom (in house, Panel TUM) (MLH1, MSH2, MSH6, PMS2, EPCAM); SNV, CNV                                                                                                                                                                                                      | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Familiärer Brust- und Eierstockkrebs (in house, Panel TUM) (BRCA1, BRCA2, ATM, CHD1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, BARD1, BRIP1, STK11, TP53); SNV, CNV                                                                                                                                                                        | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Polyposis-Syndrome (Familiäre adenomatöse Polyposis, MUTYH-assoziierte Polyposis, Cowden-Syndrom, Peutz-Jeghers-Syndrom, Hyperplastisches Polyposis-Syndrom (HPS), Serratiertes Polyposis-Syndrom (SPS), Hereditäres Mixed Polyposis-Syndrom (HMPS)) (in house, Panel TUM) (APC, MUTYH, PIK3CA, PTEN, STK11, RNF43, GREM1); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |

|                                                                                                                                                                                                                                       |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Polyposis, unbestimmt (in house, Panel TUM) (APC, MSH3, MUTYH, NTHL1, POLD1, POLE, BMPR1A, ENG, GREM1, MLH1, MSH2, MSH6, PMS2, PTEN, RNF43, SMAD4, STK11); SNV, CNV                                                                   | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Darmkrebs (CRC), unbestimmt (in house, Panel TUM) (APC, BMPR1A, ENG, EPCAM, FAN1, FLCN, GALNT12, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS1, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SETD6, SMAD4, STK11, TP53); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Gastrointestinale Stromatumore (in house, Panel TUM) (KIT, MAX, MEN1, NF1, PDGFRA, SDHAF2, SDHB, SDHC, SDHD, TMEM127)                                                                                                                 | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |

|                                                                                                                                      |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|--------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Li-Fraumeni-Syndrom (in house, Panel TUM) (TP53); SNV, CNV                                                                           | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Magenkarzinom (in house, Panel TUM) (CDH1, ATM, BMPR1A, CHEK2, MLH1, MSH2, MSH6, PMS2, STK11, TP53, CTNNA1, EPCAM, PDGFRA); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Malignes Melanom (in house, Panel TUM) (CDKN2A, BAP1, BRCA2, POT1); SNV, CNV                                                         | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |

|                                                                                                                                                                                                          |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Nierenzellkarzinom (in house, Panel TUM) (BAP1, DICER1, EPCAM, FH, FLCN, MET, PALB2, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, TSC1, TSC2, VHL); SNV, CNV                                                    | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Familiäres Pankreaskarzinom (in house, Panel TUM) (BRCA1, BRCA2, CDKN2A, CHEK2, CFTR, PALB2, STK11, APC, ATM, CFTR, MLH1, MSH2, MSH6, PALD, PMS1, PMS2, PRSS1, PTEN, RABL3, SPINK1, VHL, TP53); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Fanconi-Anämie (in house, Panel TUM) (BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, UBE2T, XRCC2); SNV, CNV                    | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |

|                                                                                                                                                                                              |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Phäochromozytome und Paragangliome / Multiple endokrine Neoplasien (in house, Panel TUM) (CDC73, CDKN1B , DLST, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Prostatakarzinom (in house, Panel TUM) (BRCA1, BRCA2, CHEK2, MSR1, RNASEL, HOXB13, PALB2, RAD51D); SNV, CNV                                                                                  | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Retinoblastom (in house, Panel TUM) (RB1); SNV, CNV                                                                                                                                          | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |

|                                                                                                                                 |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|---------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Schilddrüsenkarzinom (in house, Panel TUM) (PTEN, RET, SDHB, SDHC, SDHD, APC, ATM, CHEK2, MEN1, MUTYH, SDHAF2, STK11); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |
| Von-Hippel-Lindau-Syndrom (in house, Panel TUM) (VHL); SNV, CNV                                                                 | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 24.05.2023 |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| <p>Hereditäre Motorisch-Sensible Neuropathien/Charcot-Marie-Tooth-Neuropathien (HMSN/CMT), Distale Hereditäre Motorische Neuropathie (dHMN), Hereditäre Sensible und Autonome Neuropathien (HSAN) (in house, Panel HMSN) (AARS, AIFM1, ARHGEF10, ATL1, ATL3, ATP7A, BSCL2, CCT5, COX6A1, DCTN1, DHTKD1, DNAJB2, DNMT2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, IGHMBP2, IKBKAP, INF2, JPH1, KARS, KIF1A, KIF1B, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MORC2, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRDM12, PRPS1, PRX, RAB7, REEP1, SBF1, SBF2/MTMR13, SCN11A, SCN9A, SETX, SH3TC2, SLC12A6, SLC5A7, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, TDP1, TFG, TRIM2, TRPV4, VCP, WNK1, YARS); SNV, CNV</p> | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 14.04.2021 |
| <p>Hereditäre Neuropathie mit Neigung zu Druckpareisen (HNPP) (in house, Panel HMSN) (PMP22); SNV, CNV</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 14.04.2021 |

|                                                                                                                                                                                                                                                                                                                                                                                                                       |                       |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Kardiomyopathie, dilatativ (DCM) (in house, Panel CARDIO) (DMD, LMNA, MYBPC3, MYH7, TNNT2, TPM1, ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CASQ2, CRYAB, CSRP3, CFT1, DES, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKTN, GATAD1, ILK, LAMA2, LAMA4, LAMP2, LDB3, MURC, MYH6, MYL2, MYL3, MYPN, NEXN, NPPA, PDLIM3, PLN, PRDM16, RAF1, RBM20, SCN5A, SGCD, TAZ, TBX20, TBX5, TCAP, TMPO, TNNC1, TNNI3, TTR, VCL); SNV, CNV | Blut, DNA; DNA        | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 15.04.2019 |
| Kardiomyopathie, hypertroph (HCM) (in house, Panel CARDIO) (CADVL, ACTC1, CSRP3, FHL1, MYBPC3, MYH7, MYL2, MYL3, MYPN, PLN, PRKAG2, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, ACTN2, ANKRD1, CACNA1C, CALR3, CASQ2, CAV3, CBL, COX15 CRYAB, DES, FHL2, FXN, GLA, GAA, JPH2, KLF10, LDB3, MAP2K1, MAP2K2, MYH6, MYLK2, MYO6, MYOZ2, NEXN, PDLIM3, RAF1, RYR2, SLC25A4, TCAP, TTR, VCL); SNV, CNV                              | <u>Blut, DNA; DNA</u> | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 15.04.2019 |
| Arrhythmogene rechtsventrikuläre Kardiomyopathie / Dysplasie (ARVC/D) (in house, Panel CARDIO) (DSC2, DSG2, DSP, JUP, PKP2, TGFB3, TMEM43, CASQ2, RYR2); SNV, CNV                                                                                                                                                                                                                                                     | Blut, DNA; DNA        | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 15.04.2019 |

|                                                                                                                                     |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|-------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Non-compaction Kardiomyopathie (NCCM) (in house, Panel CARDIO) (ACTC1, HCN4, MIB1, MYBPC3, MYH7, NEXN, PRDM16, TAZ, TPM1); SNV, CNV |                |                                                                  | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 15.04.2019 |
| Kardiomyopathie, restriktiv (RCM) (in house, Panel CARDIO) (ACTC1, DES, MYH7, MYL2, MYPN, TNNT2, TNNT1); SNV, CNV                   | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA |                                                                                                                                                                                                            |                                                                                                                                                             |  | x |  | 15.04.2019 |
| Kardiomyopathie, restriktiv (RCM) (in house, Panel CARDIO) (ACTC1, DES, MYH7, MYL2, MYPN, TNNT2, TNNT1); SNV, CNV                   |                |                                                                  | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 15.04.2019 |
| Morbus Fabry (in house, Panel CARDIO) (GLA); SNV, CNV                                                                               | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 15.04.2019 |

|                                                                                                                                                                                |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Catecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) (in house, Panel CARDIO) (CASQ2, CALM1, KCNE1, KCNJ2, RYR2, TRDN); SNV, CNV                                        | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |
| Long QT-Syndrom (LQT) (in house, Panel CARDIO) (ANK2, KCNE1, KCNE2, KCNH2, KCNQ1, SCN5A, AKAP9, CACNA1C, CALM1, CAV3, KCNE3, KCNJ2, KCNJ5, RYR2, SCN4B, SNTA1, TRDN); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |
| Short QT-Syndrom (SQT) (in house, Panel CARDIO) (CACNA1C, KCNH2, KCNQ1); SNV, CNV                                                                                              | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |

|                                                                                                                                                                              |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Brugada-Syndrom (in house, Panel CARDIO) (CACNA1C, CACNA2D1, CACNB2, PKP2, SCN1B, SCN5A, TRPM4, AKAP9, CALM1, CAV3, KCNE3, KCNJ2, KCNJ5, RYR2, SCN4B, SNTA1, TRDN); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |
| Hypercholesterinämie (LDLR_Defizienz) (in house, Panel CARDIO) (LDLR); SNV, CNV                                                                                              | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |
| Hypercholesterinämie (primär) (in house, Panel CARDIO) (APOB, PCSK9, LDLRAP1); SNV                                                                                           | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline;      | AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2                                                         | Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus                                                                                       | x |  | 15.04.2019 |
| Amyloidose (erbliche Form) (in house, Panel CARDIO) (TTR); SNV                                                                                                               | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline;      | AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2                                                         | Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus                                                                                       | x |  | 15.04.2019 |

|                                                                                                                                                                                                                            |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Marfan-Syndrom (in house, Panel CARDIO) (FBN1, TGFB1, TGFB2, ACTA2, COL3A1, MYH11, MYLK, SMAD3, TGFB2); SNV                                                                                                                | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline;      | AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2                                                         | Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus                                                                                       | x |  | 15.04.2019 |
| Ehlers-Danlos-Syndrom (vaskuläre Form) (in house, Panel CARDIO) (COL3A1, ACTA2, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB1, TGFB2); SNV, CNV                                                                                   | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |
| Thorakale Aortenerweiterung (in house, Panel CARDIO) (ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB1, TGFB2); SNV, CNV                                                                                              | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |
| Bindegewebserkrankung mit Gefäßbeteiligung (in house, Panel CARDIO) (ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, LTPB2, MYH11, MYLK, MYLK2, NOTCH1, SLC2A10, SMAD3, SMAD4, TGFB2, TGFB1, TGFB2); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 15.04.2019 |

|                                                                                                                                                                            |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Maturity-onset diabetes of the young (MODY) (in house, Panel UNI) (ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1); SNV, CNV    | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 20.12.2021 |
| Familiärer Hyperinsulinismus / Hyperinsulinämische Hypoglykämie (HHF) (in house, Panel UNI) (ABCC8, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, SLC16A1, UCP2); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 20.12.2021 |
| Familiäre Hypercholesterinämie (FH) (in house, Panel UNI) (APOB, LDLR, LDLRAP1, PCSK9); SNV, CNV                                                                           | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 20.12.2021 |

|                                                                                                                                                                                                                                                                                                                |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Familiäre hypokalzurische Hyperkalzämie (HHC) (in house, Panel UNI) (CASR, AP2S1, GNA11); SNV, CNV                                                                                                                                                                                                             | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 20.12.2021 |
| Kallmann-Syndrom / Isolierter Hypogonadotroper Hypogonadismus (IHH) (in house, Panel UNI) (ANOS1/KAL1, AXL, CCDC141, CHD7, DUSP6, FEZF1, FGF8, FGF17, FGFR1, FLRT3, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, NSMF, POLR3B, PROK2, PROKR2, TAC3, TACR3, SEMA3A, SOX10, SPRY4, SRA1, WDR11); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 20.12.2021 |
| Morbus Osler / Hereditäre Hämorrhagische Teleangiectasie (HHT) (in house, Panel UNI) (ACVRL1, ENG, SMAD4, GDF2, RASA1); SNV, CNV                                                                                                                                                                               | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 20.12.2021 |

|                                                                                                                                                                                                                                                                                                                      |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Cystische Fibrose (CF) (in house, Panel UNI) CFTR                                                                                                                                                                                                                                                                    |                |                                                                  | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 20.12.2021 |
|                                                                                                                                                                                                                                                                                                                      | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
| Retinitis pigmentosa, autosomal-dominant und x-gekoppelt (in house, Panel AUGE) (ARL3, BEST1, CA4, CACNA1F, CRX, FSCN2, GUCA1B, HK1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, OFD1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, SPP2, TOPORS); SNV, CNV |                |                                                                  | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |
|                                                                                                                                                                                                                                                                                                                      | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Retinitis pigmentosa, autosomal-rezessiv und x-gekoppelt (in house, Panel AUGÉ) (ABCA4, ADGRA3, ADIPOR1, AGBL5, AHI1, AIPL1, ARHGEF18, ARL2BP, ARL6, BBS2, C8ORF37, CDHR1, CEP290, CERKL, CLCC1, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CWC27, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, GNAT1, GNPTG, GUCY2D, HGSNAT, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIF11, KIZ, LRAT, MAK, MERTK, MFRP, NEK2, NEUROD1, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF31, RBP3, RCBTB1, RDH11, RDH12, REEP6, RGR, RHO, RLBP1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, SAG, SAMD11, SLC7A14, SPATA7, TRNT1, TTC8, TUB, TULP1, USH2A, WDR19, ZNF408, ZNF513); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |
| ROSAH-Syndrom (in house, Panel AUGÉ) (ALPK1); SNV, CNV                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                         |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Mikrocornea, myopische chorioretinale Atrophy, Telecanthus (in house, Panel AUGE) ADAMTS18                                                                                                                                              | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Usher-Syndrom (in house, Panel AUGE) (ABHD12, ADGRV1, ARSG, CDH23, CEP250, CIB2, CLRN1, GJB2, HARS, MYO7A, PCDH15, PDZD7, PRPH2, USH1C, USH1G, USH2A, WHRN); SNV, CNV                                                                   | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Refsum-Syndrom / Zellweger-Syndrom-Spektrum (Refsum / Zellweger / Neonatale Adrenoleukodystrophie) (in house, Panel AUGE) (PHYH, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                                                                                    |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Bardet-Biedl-Syndrom (in house, Panel AUGE) (ARL6, BBIP, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CCDC28B, CEP290, IFT27, IFT74, IFT172, INPP5E, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC8, TTC21B, WDPCP); SNV, CNV                                              | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Alagille-Syndrom (in house, Panel AUGE) (JAG1, NOTCH2, BMP2); SNV, CNV                                                                                                                                                                                                                             | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Leber'sche kongenitale Amaurose (in house, Panel AUGE) (AGBL5, AIPL1, ALMS1, CABP4, CDH16, CEP290, CLUAP1, CNGA3, CRB1, CRX, DTHD1, GDF6, GUCY2D, IDH3A, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, NMNAT1, OTX2, PDE6G, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Kongenitale stationäre Nachtblindheit (in house, Panel AUGE) (CABP4, CACNA1F, CACNA2D4, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1); SNV, CNV                                                                                                                                                                                                                                               | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Zapfendystrophie (Zapfen-Stäbchen-Dystrophie) (in house, Panel AUGE) (ABCA4, ADAM9, AIPL1, ALMS1, ATF6, BEST1, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP78, CEP250, CERKL, CFAP410, CNGA3, CNGB3, CNNM4, CRB1, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, NMNAT1, NR2E3, NRL, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RDH12, RGS9, RGS9BP, RIMS1, RPGR (inkl. ORF15), RPGRIP1, SEMA4A, TTLL5, TULP1, UNC119); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Makuladystrophien / Morbus Stargardt (in house, Panel AUGE) (ABCA4, BEST1, C1QTNF5, CDH3, CFH, CLN3, CNGB3, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, GUCA1A, GUCY2D, IMPG1, IMPG2, IRX1, MAPKAP3, MFSD8, PPT1, PRDM13, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTLL5); SNV, CNV                                                                                                                                                                                     | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                                                                                          |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Optikusatrophie und Leber´sche hereditäre Optikusneuropathie (LHON) (in house, Panel AUGÉ) (MT-ND1, MT-ND4, MT-ND4L, MT-ND6, ACO2, AFG3L2, ANTXR1, ATP1A3, C12ORF65, CISD2, DNM1L, FDXR, MFN2, MTPAP, NBAS, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, WFS1, YME1L1); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Flecked Retina Erkrankungen (in house, Panel AUGÉ) (ABCA4, CHM, CYP4V2, EFEMP1, ELOVL4, KCNJ13, OAT, PLA2G5, PROM1, PRPH2, RDH5, RHO, RLBP1, RPE65, RS1, VPS13B); SNV, CNV                                                                                                                               | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Vitreoretinopathien (Familiäre Exsudative Vitreoretinopathie) (in house, Panel AUGÉ) (ATOH7, BEST1, CAPN5, COL18A1, COL2A1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RCBTB1, TSPAN12, VCAN, ZNF408); SNV, CNV                                                                                | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |

|                                                                                                                                                                                   |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Senior-Loken-Syndrom (in house, Panel AUGE) (CEP164, CEP290, INVS, IFT81, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TMEM67, TRAF3IP1, WDR19, ZNF423); SNV, CNV                         | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |
| Achromatopsie (in house, Panel AUGE) (ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H); SNV, CNV                                                                                          | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |
| Myopie und Differentialdiagnosen (in house, Panel AUGE) (COL11A1, COL11A2, COL18A1, COL2A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, FBN1, FZD4, LRP2, LRP5, P3H2, VCAN); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Katarakt (in house, Panel AUGÉ)<br>(ABCB6, ABHD12, ADAMTSL4, AGK, BFSP1, BFSP2, CHMP4B, CLPB, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP51A1, CYP27A1, DNMBP, EPG5, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, GALK1, GALT, GCNT2, GEMIN4, GJA3, GJA8, GLA, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, LTBP2, MAF, MIP, MIR184, MYH9, NDP, NHS, OCRL, OPA3, P3H2, PAX6, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RECQL2, RECQL4, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TMEM114, UNC45B, VIM, VSX2, WFS1); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |
| Mikrophthalmie-Anophthalmie-Kolobom-Komplex (in house, Panel AUGÉ)<br>(ABCB6, ACTB, ACTG1, ATOH7, C12ORF57, CRYBA4, CYP1B1, ERCC1, ERCC2, ERCC5, ERCC6, FOXE3, FOXL2, FRAS1, FREM2, FZD4, GDF3, GDF6, GJA1, GRIP1, HESX1, HMX1, MFRP, NDUFB11, OCRL, OTX2, PAX2, PAX6, PIGL, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RAX, RBP4, SIX3, SIX6, SOX2, PXDN, RAB18, RAB3GAP1, RAB3GAP2, TMEM98, VPS13B, VSX2, ZIC2); SNV, CNV                                                                                                                                                                                                                                                                     | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                                                      |                |                                                                     |                                                                                                                                                                                                                                           |                                                                                                                                                                                              |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|---------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Linsenluxation (in house, Panel AUGE)<br>(ADAMTS10, ADAMTS17, ADAMTSL4,<br>ASPH, CBS, COL11A1, COL18A1,<br>COL2A1, FBN1, LTBP2, P3H2, VCAN,<br>VSX2); SNV, CNV                                                                                                       | Blut, DNA; DNA | Sequence capture; Sequencing-by<br>synthesis; custom pipeline; MLPA | AA-112, V006; AA-113,<br>V005; AA-156, V004; AA-<br>157, V003; AA-209,<br>V003; AA-247, V005; AA-<br>501, V003; AA-502,<br>V002; AA-504, V004; AA-<br>506, V006; AA-509,<br>V003; AA-510, V002; AA-<br>511, V002; Varvis V2.73;<br>ICA V2 | ThermoFisher<br>Scientific,<br>SeqStudio;<br>ThermoFisher<br>Scientific, Genetic<br>Analyzer 3500xl;<br>Illumina,<br>Nextseq550;<br>Illumina,<br>NovaSeq6000;<br>Illumina, NovaSeq<br>X Plus | x |  | 06.11.2023 |
| Hornhautdystrophien/Keratoconus (in<br>house, Panel AUGE) (AGBL1, CHST6,<br>COL17A1, COL8A2, CYP4V2, DCN,<br>GSN, KRT3, KRT12, LOXHD1,<br>MIR184, OVOL2, PAX6, PIKFYVE,<br>PRDM5, SLC4A11, TACSTD2, TCF4,<br>TGFB1, TUBA3D, UBIAD1, VSX1,<br>ZEB1, ZNF469); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by<br>synthesis; custom pipeline; MLPA | AA-112, V006; AA-113,<br>V005; AA-156, V004; AA-<br>157, V003; AA-209,<br>V003; AA-247, V005; AA-<br>501, V003; AA-502,<br>V002; AA-504, V004; AA-<br>506, V006; AA-509,<br>V003; AA-510, V002; AA-<br>511, V002; Varvis V2.73;<br>ICA V2 | ThermoFisher<br>Scientific,<br>SeqStudio;<br>ThermoFisher<br>Scientific, Genetic<br>Analyzer 3500xl;<br>Illumina,<br>Nextseq550;<br>Illumina,<br>NovaSeq6000;<br>Illumina, NovaSeq<br>X Plus | x |  | 06.11.2023 |
| Septo-optische Dysplasie (in house,<br>Panel AUGE) (FGFR1, HESX1, OTX2,<br>PROKR2, SOX2, SOX3); SNV, CNV                                                                                                                                                             | Blut, DNA; DNA | Sequence capture; Sequencing-by<br>synthesis; custom pipeline; MLPA | AA-112, V006; AA-113,<br>V005; AA-156, V004; AA-<br>157, V003; AA-209,<br>V003; AA-247, V005; AA-<br>501, V003; AA-502,<br>V002; AA-504, V004; AA-<br>506, V006; AA-509,<br>V003; AA-510, V002; AA-<br>511, V002; Varvis V2.73;<br>ICA V2 | ThermoFisher<br>Scientific,<br>SeqStudio;<br>ThermoFisher<br>Scientific, Genetic<br>Analyzer 3500xl;<br>Illumina,<br>Nextseq550;<br>Illumina,<br>NovaSeq6000;<br>Illumina, NovaSeq<br>X Plus | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                                                             |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Kongenitales Glaukom / Axenfeld-Rieger-Syndrom (in house, Panel AUGÉ)<br>(ASB10, COL4A1, CYP1B1, DDX58, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, SBF2, SH3PXD2B, TBK1, TEK, WDR36); SNV, CNV                                                     | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Vorderkammerdysgenesien / Axenfeld-Rieger-Syndrom (in house, Panel AUGÉ) (ASPH, B3GLCT, COL4A1, COL4A2, CYP1B1, EYA1, FOXC1, FOXE3, GPATCH3, HMX1, LTBP2, MYOC, PAX6, PITX2, PXDN, SLC38A8); SNV, CNV                                                                       | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |
| Albinismus (syndromal) (in house, Panel AUGÉ) (AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, EDN3, EDNRB, EPG5, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC38A8, SLC45A2, SOX10, TYR, TYRP1); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 06.11.2023 |

|                                                                                                                                                                                                                                                                                                                                                                         |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Ichtyosis (nicht syndromal/syndromal) (in house, Panel HAUT) (FLG, STS, ABCA12, ALOX12B, ALOXE3, CDSN, CERS3, CHST8, CSTA, CYP4F22, FLG2, GJB3, GJB4, KRT1, KRT10, KRT2, LIPN, NIPAL4, PNPLA1, SDR9C7, SERPINB8, ST14, SULT2B1, TGM1, TGM5, ABHD5, ALDH3A2, AP1B1, AP1S1, CLDN1, EBP, ELOVL4, GJB2, MBTPS2, NSDHL, PHYH, PIGL, SLC27A4, SNAP29, SPINK5, ST14); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 19.10.2022 |
| Ektodermale Dysplasien (in house, Panel HAUT) (CDH3, CST6, DLX3, EDA, EDAR, EDARADD, GJA1, GJB6, HOXC13, IFT122, IFT43, IKBKG, KDF1, KREMEN1, KRT74, KRT85, MSX1, NECTIN1, NECTIN4, NFKBIA, PORCN, TP63, TSPEAR, WDR19, WDR35, WNT10A); SNV, CNV                                                                                                                        | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 19.10.2022 |
| Epidermolysis bullosa (in house, Panel HAUT) (CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSP, DST, EXPH5, FLG2, ITGA3, ITGA6, ITGB4, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1, PLEC1, SERPINB8, TGM5); SNV, CNV                                                                                                                                      | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 19.10.2022 |

|                                                                                                                                                                                                                                                                    |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Tumorerkrankungen der Haut (Gorlin-Goltz-Syndrom, Malignes Melanom, Birt-Hogg-Dube-Syndrom, Familiäre Leiomyomatose mit Nierenzellkarzinom) (in house, Panel HAUT) (PTCH1, PTCH2, SUFU, CDK4, CDKN2A, MC1R, MITF, POT1, TYR, FLCN, FH); SNV, CNV                   | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |
| Bindegewebserkrankungen (Marfan-Syndrom, Cutis laxa, Pseudoxanthoma elasticum) (in house, Panel HAUT) (CBS, FBN1, FBN2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, EFEMP2, ELN, FBLN5, LTBP4, PYCR1, ABCC6, XYLT1, XYLT2); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |
| Neurokutane Syndrome (Neurofibromatose, Tuberöse Sklerose) (in house, Panel HAUT) (LZTR1, NF1, NF2, SMARCB1, SPRED1, TSC1, TSC2); SNV, CNV                                                                                                                         | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|---|--|------------|
| Acne inversa (in house, Panel HAUT) (PSENEN, NCSTN, PSEN1); SNV, CNV                                                                                                                                                                                                                                                                                                                                                                                                                                           | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 19.10.2022 |
| Pigmentierungsstörungen (Incontinentia pigmenti, oculokutaner Albinismus, syndromale Hypopigmentierungsstörungen, Morbus Dowling-Degos und andere Hyperpigmentierungsstörungen) (in house, Panel HAUT) (IKBKG, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1, AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LYST, MITF, MLPH, MYO5A, PAX3, RAB27A, SNAI2, SOX10, ABCB6, ADAM10, ADAR, KITLG, KRT14, KRT5, POFUT1, POGLUT1, POLA1, PSENEN, SASH1); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 19.10.2022 |
| Hereditäres Angioödem (in house, Panel HAUT) (C1NH, F12); SNV, CNV                                                                                                                                                                                                                                                                                                                                                                                                                                             | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus | x |  | 19.10.2022 |

|                                                                                                                                                                                                                                                                            |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Hereditäres Lymphödem (in house, Panel HAUT) (CCBE1, FAT4, FLT4, FOXC2, GJC2, PIEZO1, VEGFC); SNV, CNV                                                                                                                                                                     | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |
| Palmoplantare Keratosen (in house, Panel HAUT) (AAGAB, AQP5, CAST, CTSB, CTSC, DSG1, DSP, FERMT1, GJB2, JUP, KANK2, KRT1, KRT10, KRT14, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT9, LOR, MBTPS2, PKP1, POMP, RHBDF2, RSPO1, SERPINB7, SLURP1, SMARCAD1, TAT, TRPV3); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |
| Follikuläre Verhornungsstörungen (in house, Panel HAUT) (ATP2A2, ATP2C1, MBTPS2, RIPK4); SNV, CNV                                                                                                                                                                          | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |

|                                                                                                                                                                                     |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| Trichothiodystrophie (in house, Panel HAUT) (ERCC2, ERCC3, GTF2E2, GTF2H5, MPLKIP, RNF113A, TARS1); SNV, CNV                                                                        | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |
| Haaranomalien (in house, Panel HAUT) (ANTXR1, APCDD1, CDH3, CDSN, DSG4, DSP, EPS8L3, HR, KRT71, KRT74, KRT81, KRT83, KRT86, LIPH, LPAR6, LSS, POC1A, RPL21, SNRPE, SOX18); SNV, CNV | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |
| Nageldysplasien (in house, Panel HAUT) (COL7A1, EDAR, FZD6, HOXC13, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT74, KRT85, PLCD1, RIPK4, RSPO4); SNV, CNV                                 | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 19.10.2022 |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |                |                                                                  |                                                                                                                                                                                                            |                                                                                                                                                             |  |   |  |            |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| <p>Erbliche Hörstörungen (in house, Panel DFN) (ACTG1, ADCY1, ADGRV1, AIFM1, ATP2B2, ATP6B1 (ATP6V1B1), CABP2, CACNA1D, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, CLRN1-AS1, COCH, COL11A1, COL11A2, COL4A3, COL4A4, COL4A5, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GRHL2, GRXCR1, GRXCR2, GSDME (DFNA5), HARS, HGF, HOMER2, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MCM2, MET, MIR96, MITF, MSRB3, MTCO1, MTND1, MTRNR1, MTTH, MTTI, MTTL1, MTT51, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX6,</p> | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V006; AA-113, V005; AA-156, V004; AA-157, V003; AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  | 20.12.2021 |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |                |                                                                  |                                                                                                                                                                                |                                                                                                                                                             |  |   |  |            |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---|--|------------|
| <p>PJVK, PNPT1, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, ROR1, S1PR2, SANS (USH1G), SERPINB6, SIX1, SIX5, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC44A4, SLITRK6, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TECTA, TECTA, TIMM8A, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, USH2A, WBP2, WFS1, WHRN, WS2B, RIMBP2, ATP1A3, LRBA, ATP11A, PPIP5K2, SYNJ2, CEP78, MRPS2, TD, PHYH, PEX7, ARSG, ABCA4, ABHD12, ALMS1, ANKH, BCAP31, BCS1L, BDP1, BSND, C10ORF2, CATSPER2, CD151, CDKN1C, CHD7, CHSY1, COL2A1, COL9A1, COL9A2, DNMT1, GPRASP2, GPSM2, HARS2, HOXB1, HSD17B4, LARS2, MANBA, NDP, NLRP3, POLR1C, POLR1D, RPE65, SALL1, SEMA3E, SLC19A2, SPATA5, TCOF1, TFAP2A, TJP2, TMC2, TYR, KARS1, LARS2, TUBB4B); SNV, CNV</p> | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA | AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2 | ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus |  | x |  |            |
| <p>Erblich bedingte mitochondriale Erkrankungen (in house, Panel mtDNA) (MT-TV, MT-RNR2, MT-TL1, MT-ND1, MT-TI, MT-TQ, MT-TM, MT-ND2, MT-TW, MT-TA, MT-TN, MT-TC, MT-TY, MT-CO1, MT-TS1, MT-TD, MT-CO2, MT-TK, MT-ATP8, MT-ATP6, MT-CO3, MT-TG, MT-ND3, MT-TR, MT-NDL4, MT-ND4, MT-TH, MT-TS2, MT-TL2, MT-ND5, MT-ND6, MT-TE, MT-CYB, MT-TT, MT-TP); SNV</p>                                                                                                                                                                                                                                                                                                                                                                                 | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline;      | AA-209, V003; AA-247, V005; AA-501, V003; AA-502, V002; AA-504, V004; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2                             | Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus                                                                                       |  | x |  | 21.04.2023 |
| <p>Whole Exome Sequencing (WES); SNV, CNV (Agilent SureSelect Human All Exon V8)</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | Blut, DNA; DNA | Sequence capture; Sequencing-by synthesis; custom pipeline;      | AA-209, V003; AA-247, V005; AA-501, V003; AA-504, V004; AA-505, V005; AA-506, V006; AA-509, V003; AA-510, V002; AA-511, V002; Varvis V2.73; ICA V2                             | Illumina, Nextseq550; Illumina, NovaSeq6000; Illumina, NovaSeq X Plus                                                                                       |  | x |  | 20.12.2021 |

|                                                                                |                |                                                                |                                                                                                                        |                                                          |  |   |  |  |            |
|--------------------------------------------------------------------------------|----------------|----------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------|--|---|--|--|------------|
| Whole Genome Sequencing (WGS);<br>SNV, CNV, SV (Illumina DNA PCR-Free<br>Prep) | Blut, DNA; DNA | Sequence capture; Sequencing-by<br>synthesis; custom pipeline; | AA-209, V003; AA-501,<br>V002; AA-505, V005; AA-<br>506, V006; AA-508,<br>V003; AA-510, V002; AA-<br>511, V002; ICA V2 | Illumina,<br>NovaSeq6000;<br>Illumina, NovaSeq<br>X Plus |  | x |  |  | 26.07.2024 |
|--------------------------------------------------------------------------------|----------------|----------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------|--|---|--|--|------------|