



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

**Untersuchungsart:
 Chromosomenanalyse****

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	Gerät	CE-Verfahren	in Haus-Verfahren	Erläuterung zu weiterer Bearbeitung, sofern zutreffend	Datum der Aufnahme/Änderung
Angeborener Chromosomensatz	peripheres Blut, Fruchtwasser	Chromosomenbänderungsanalyse	AA-201, V005; AA-206, V004	Zeiss, Axio Imager, Metasystems, Ikaros		x		22.01.2013
Angeborener Chromosomensatz	Fruchtwasser	Fluoreszenz-in-situ-Hybridisierung mit spezifischen Sonden (FISH)	AA-205, V003	Zeiss, Axio Imager, Metasystems, Isis	x			22.01.2013

**Untersuchungsart:
 Molekularbiologische Untersuchungen (Amplifikationsverfahren)****

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	Gerät	CE-Verfahren	in Haus-Verfahren	Erläuterung zu weiterer Bearbeitung, sofern zutreffend	Datum der Aufnahme/Änderung
Fragiles X-Syndrom (FMR1)	Blut, DNA; DNA	Fragmentlängenanalyse	AA-103, V004; AA-156, V004; AA-157, V003; AA-209, V003; AA-248, V003; AA-249, V003; AA-250, V004	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic 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-polyposes 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 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		24.05.2023
Familiäres Pankreaskarzinom (in house, Panel TUM) (BRCA1, BRCA2, CDKN2A, CHEK2, CFTR, PALB2, STK11, APC, ATM, CFTR, MLH1, MSH2, MSH6, PALLD, 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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>14.04.2021</p>
<p>Hereditäre Neuropathie mit Neigung zu Druckpareesen (HNPP) (in house, Panel HMSN) (PMP22); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>14.04.2021</p>

<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>15.04.2019</p>
<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>15.04.2019</p>
<p>Arrhythmogene rechtsventrikuläre Kardiomyopathie / Dysplasie (ARVC/D) (in house, Panel CARDIO) (DSC2, DSG2, DSP, JUP, PKP2, TGFB3, TMEM43, CASQ2, RYR2); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>15.04.2019</p>

Non-compaction Kardiomyopathie (NCCM) (in house, Panel CARDIO) (ACTC1, HCN4, MIB1, MYBPC3, MYH7, NEXN, PRDM16, TAZ, TPM1); 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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		15.04.2019
Kardiomyopathie, restriktiv (RCM) (in house, Panel CARDIO) (ACTC1, DES, MYH7, MYL2, MYPN, TNNI3, TNNT2, TPM1); 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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		15.04.2019
Hypercholestrinämie (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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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		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		x		15.04.2019
Marfan-Syndrom (in house, Panel CARDIO) (FBN1, TGFBR1, TGFBR2, 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		x		15.04.2019
Ehlers-Danlos-Syndrom (vaskuläre Form) (in house, Panel CARDIO) (COL3A1, ACTA2, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2); 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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Thorakale Aortenerweiterung (in house, Panel CARDIO) (ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2); 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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019

<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>15.04.2019</p>
<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>20.12.2021</p>
<p>Familiärer Hyperinsulinismus / Hyperinsulinämische Hypoglykämie (HHF) (in house, Panel UNI) (ABCC8, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, SLC16A1, UCP2); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>20.12.2021</p>
<p>Familiäre Hypercholesterinämie (FH) (in house, Panel UNI) (APOB, LDLR, LDLRAP1, PCSK9); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>20.12.2021</p>

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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		20.12.2021
Cystische Fibrose (CF) (in house, Panel UNI) CFTR	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		20.12.2021

<p>Retinitis pigmentosa, autosomal-dominant und x-gekoppelt (in house, Panel AUGÉ) (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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>ROSAH-Syndrom (in house, Panel AUGÉ) (ALPK1); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>

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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		06.11.2023

<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>Optikusatrophie und Leber´sche hereditäre Optikusneuropathie (LHON) (in house, Panel AUGE) (MT-ND1, MT-ND4, MT-ND4L, MT-ND6, ACO2, AFG3L2, ANTXR1, ATP1A3, C12ORF65, CISD2, DNMT1L, FDXR, MFN2, MTPAP, NBAS, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, WFS1, YME1L1); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>

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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		06.11.2023
Vitreoretinopathien (Familiäre Exsudative Vitreoretinopathie) (in house, Panel AUGÉ) (ATOH7, BEST1, CAPN5, COL18A1, COL2A1, CTNNA1, 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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		06.11.2023
Senior-Loken-Syndrom (in house, Panel AUGÉ) (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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		06.11.2023
Achromatopsie (in house, Panel AUGÉ) (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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		06.11.2023

<p>Myopie und Differentialdiagnosen (in house, Panel AUGÉ) (COL11A1, COL11A2, COL18A1, COL2A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, FBN1, FZD4, LRP2, LRP5, P3H2, VCAN); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>			<p>06.11.2023</p>
<p>Katarakt (in house, Panel AUGÉ) (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, CTD1P1, 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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>			<p>06.11.2023</p>

<p>Mikrophthalmie-Anophthalmie-Kolobom-Komplex (in house, Panel AUGE) (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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>Linsenluxation (in house, Panel AUGE) (ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, CBS, COL11A1, COL18A1, COL2A1, FBN1, LTBP2, P3H2, VCAN, VSX2); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>Hornhautdystrophien/Keratoconus (in house, Panel AUGE) (AGBL1, CHST6, COL17A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, MIR184, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>

<p>Septo-optische Dysplasie (in house, Panel AUGE) (FGFR1, HESX1, OTX2, PROKR2, SOX2, SOX3); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>Kongenitales Glaukom / Axenfeld-Rieger-Syndrom (in house, Panel AUGE) (ASB10, COL4A1, CYP1B1, DDX58, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, SBF2, SH3PXD2B, TBK1, TEK, WDR36); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>Vorderkammerdysgenesien / Axenfeld-Rieger-Syndrom (in house, Panel AUGE) (ASPH, B3GLCT, COL4A1, COL4A2, CYP1B1, EYA1, FOXC1, FOXE3, GPATCH3, HMX1, LTBP2, MYOC, PAX6, PITX2, PXDN, SLC38A8); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>
<p>Albinismus (syndromal) (in house, Panel AUGE) (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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>

<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>
<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>
<p>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</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>
<p>Tumorerkrankungen der Haut (Gorlin-Goltz-Syndrom, Malignes Melanom, Birt-Hogg-Dube-Syndrom, Familiäre Leiomyomatose mit Nirenzellkarzinom) (in house, Panel HAUT) (PTCH1, PTCH2, SUFU, CDK4, CDKN2A, MC1R, MITF, POT1, TYR, FLCN, FH); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>

<p>Bindegeweberkrankungen (Marfan-Syndrom, Cutis laxa, Pseudoxanthoma elasticum) (in house, Panel HAUT) (CBS, FBN1, FBN2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, EFEMP2, ELN, FBLN5, LTBP4, PYCR1, ABCC6, XYLT1, XYLT2); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>			<p>19.10.2022</p>
<p>Neurokutane Syndrome (Neurofibromatose, Tuberöse Sklerose) (in house, Panel HAUT) (LZTR1, NF1, NF2, SMARCB1, SPRED1, TSC1, TSC2); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>			<p>19.10.2022</p>
<p>Acne inversa (in house, Panel HAUT) (PSENEN, NCSTN, PSEN1); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>			<p>19.10.2022</p>

<p>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, PSENN, SASH1); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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; Varis V2.73; ICA V2</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>
<p>Hereditäres Angioödem (in house, Panel HAUT) (C1NH, F12); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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; Varis V2.73; ICA V2</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>
<p>Hereditäres Lymphödem (in house, Panel HAUT) (CCBE1, FAT4, FLT4, FOXC2, GJC2, PIEZO1, VEGFC); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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; Varis V2.73; ICA V2</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>

Palmoplantare Keratosen (in house, Panel HAUT) (AAGAB, AQP5, CAST, CTSC, 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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	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 3500xIllumina, Nextseq550; Illumina, NovaSeq6000	x		19.10.2022

<p>Nageldysplasien (in house, Panel HAUT) (COL7A1, EDAR, FZD6, HOXC13, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT74, KRT85, PLCD1, RIPK4, RSPO4); SNV, CNV</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>19.10.2022</p>
<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, MTTs1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX6,</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>20.12.2021</p>

<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>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA</p>	<p>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</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xIllumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		
<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>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline;</p>	<p>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</p>	<p>Illumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>21.04.2023</p>
<p>Whole Exome Sequencing (WES); SNV, CNV (Agilent SureSelect Human All Exon V8)</p>	<p>Blut, DNA; DNA</p>	<p>Sequence capture; Sequencing-by synthesis; custom pipeline;</p>	<p>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</p>	<p>Illumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>20.12.2021</p>

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