



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

Untersuchungsart:

Chromosomenanalyse ^[Flex C]

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) ^[Flex C]

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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000</p>	<p>x</p>		<p>06.11.2023</p>

Flecked Retina Erkrankungen (in house, Panel AUGE) (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	x		06.11.2023
Vitreoretinopathien (Familiäre Exsudative Vitreoretinopathie) (in house, Panel AUGE) (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 3500xl; Illumina, Nextseq550; Illumina, NovaSeq6000	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	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	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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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; Varvis V2.73; ICA V2</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, 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; Varvis V2.73; ICA V2</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, 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; Varvis V2.73; ICA V2</p>	<p>ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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 3500xl; Illumina, 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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