Universitätsmedizin Göttingen
Institut für Humangenetik
Heinrich-Düker-Weg 12
37073 Göttingen

Untersuchungsverfahren

Dokument ID: 145229
Stand: 31.10.2025

Untersuchungsgebiet: Humangenetik (Zytogenetik)

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, Fibroblasten, Fruchtwasser	Chromosomenbänderungsanalyse	AA-201, V004; AA-206, V003	Zeiss, Axio Imager, Metasystems, Ikaros		x		22.01.2013
Angeborener Chromosomensatz	Fruchtwasser	Fluoreszenz-in-situ-Hybridisierung mit spezifischen Sonden (FISH)	AA-205, V002	Zeiss, Axio Imager, Metasystems, Isis	x			22.01.2013
Angeborener Chromosomensatz	kultivierte Amnionzellen, peripheres Blut; DNA	Molekulare Karyotypisierung mittels Array-CGH	AA-114, V002; AA-209, V003	Agilent, SureScan Dx G5761A Microarray Scanner		x		22.01.2013

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

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, V003; AA-156, V003; AA-157, V002; AA-209, V003; AA-248, V002; AA-249, V002; AA-250, V003	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, V003; AA-156, V003; AA-157, V002; AA-209, V003; AA-252, V002	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl		x		13.12.2022
Chorea Huntington (HD) (HTT)	Blut, DNA; DNA	Fragmentlängenanalyse	AA-103, V003; AA-156, V003; AA-157, V002; AA-209, V003; AA-254, V002	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl		x		13.12.2022
Friedreichsche Ataxie (FRDA) (FXN)	Blut, DNA; DNA	Fragmentlängenanalyse	AA-103, V003; AA-156, V003; AA-157, V002; AA-209, V003; AA-253, V002	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl		x		10.02.2023
Prader-Willi-Syndrom/Angelman-Syndrom (UBE3A)	Blut, DNA; DNA	MS-MLPA	AA-113, V004; AA-153, V003; AA-156, V003; AA-157, V002; AA-209, V003;	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl		x		29.08.2018
Fragiles-X-Syndrom (FMR1)	Blut, DNA; DNA	MS-MLPA	AA-113, V004; AA-153, V003; AA-156, V003; AA-157, V002; AA-209, V003;	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl		x		27.05.2020

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Beckwith-Wiedemann-Syndrom / Silver-Russell-Syndrom (ICR1, ICR2)	Blut, DNA; DNA	MS-MLPA	AA-113, V004; AA-153, V003; AA-156, V003; AA-157, V002; AA-209, V003;	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xl		x		20.11.2018
Lynch-Syndrom / Hereditäres Nicht-polyposes Kolorektalkarzinom (Panel TUM) (MLH1, MSH2, MSH6, PMS2, EPCAM); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Familiärer Brust- und Eierstockkrebs (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, 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)) (Panel TUM) (APC, MUTYH, PIK3CA, PTEN, STK11, RNF43, GREM1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Polyposis, unbestimmt (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Darmkrebs (CRC), unbestimmt (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Gastrointestinale Stromatumore (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Li-Fraumeni-Syndrom (Panel TUM) (TP53); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Magenkarzinom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Malignes Melanom (Panel TUM) (CDKN2A, BAP1, BRCA2, POT1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023

Nierenzellkarzinom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Familiäres Pankreaskarzinom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Fanconi-Anämie (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Phäochromozytome und Paragangliome / Multiple endokrine Neoplasien (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Prostatakarzinom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Retinoblastom (Panel TUM) (RB1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Schilddrüsenkarzinom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Von-Hippel-Lindau-Syndrom (Panel TUM) (VHL); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Makrozephalie (Basisdiagnostik) (Panel MAZ) (CHD8, DNMT3A, EZH2, NSD1, NFIX, PTEN); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Weaver-Syndrom (Panel MAZ) (EZH2, EED, SUZ12, HACE1, NSD1, NFIX); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019

Sotos-Syndrom (Panel MAZ) (NSD1, NFIX, EZH2, EED, SUZ12, HACE1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Beckwith-Wiedemann-Syndrom (Panel MAZ) (H19, KCNQ1OT1, CDKN1C, NSD1, ICR1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Alexander-Krankheit (Panel MAZ) (GFAP); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Bannayan-Riley-Ruvalcaba-Syndrom, Lhermitte-Duclos-Syndrom, Cowden syndrome 1, Macrozephalie/Autismus-Syndrom (Panel MAZ) (PTEN); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Brittle Cornea-Syndrom (Panel MAZ) (ZNF469); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Dysplasie, kranio-metaphysäre (CMD) (Panel MAZ) (ANKH, GJA1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Greig-Zephalopolysyndaktylie-Syndrom (Panel MAZ) (GLI3, KIF7); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Joubert-Syndrom 12, Akrokallosales Syndrom (Panel MAZ) (KIF7); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Hydrozephalus mit Morbus Hirschsprung (Panel MAZ) (L1CAM); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Kosaki-Syndrom (Panel MAZ) (PDGFRB); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Krämpfe-Skoliose-Makrozephalie –Syndrom (SSMS) (Panel MAZ) (EXT2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019

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Untersuchungsverfahren

Luscan-Lumish-Syndrom (LLS) (Panel MAZ) (SETD2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
MACS-Syndrom / RIN2-Syndrom (Panel MAZ) (RIN2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Makrozephalie mit Makrosomie und Gesichtsdysmorphien (MMFD) (Panel MAZ) (RNF135); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Megalenzephalie-Polymikrogyrie-Polydaktylie-Hydrozephalus-Syndrom (MPPH1-3) (Panel MAZ) (PIK3R2, AKT3, CCND2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Megalenzephalen Leukoenzephalopathie mit subkortikalen Zysten (MLC) (Panel MAZ) (HEPACAM, MLC1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Megalenzephalie-Kapillar-Malformation-Polymikrogyrie-Syndrom (MCAP) (Panel MAZ) (PIC3CA); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Mentale Retardierung-Makrozephalie, autosomal rezessiv (MRT41) (Panel MAZ) (KPTN); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Mentale Retardierung-Makrozephalie, Suszeptibilität für Autismus (AUTS18) (Panel MAZ) (CHD8); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Mentale Retardierung-Makrozephalie, X-chromosomal (Cabezas; MRX93; MRX72; Turner type) (Panel MAZ) (CUL4B, BRWD3, RAB39B, HUWE1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Opitz-Kaveggia-Syndrom (OKS) (Panel MAZ) (MED12); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Perlman-Syndrom (PRLNMS) (Panel MAZ) (DIS3L2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019

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Phelan-McDermid-Syndrom (Panel MAZ) (SHANK3); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Polyhydramnion, Megalenzephalie und Symptomatische Epilepsie (Panel MAZ) (STRADA); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Proteus-Syndrom (Panel MAZ) (AKT1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Simpson-Golabi-Behmel-Syndrom (SGBS) (Panel MAZ) (GPC3, OFD1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Smith-Kingsmore-Syndrom (SKS) (Panel MAZ) (MTOR); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Tatton-Brown-Rahman-Syndrom (TBRS) (Panel MAZ) (DNMT3A)	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Tenorio-Syndrom (TNORS) (Panel MAZ) (RNF125); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019
Makrozephalie- Gesamt-Diagnostik (Panel MAZ) (AKT1, AKT3, ANKH, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DIS3L2, DNMT3A, EED, EXT2, EZH2, GFAP, GJA1, GLI3, GPC3, H19, HACE1, HEPACAM, HUWE1, KCNQ1OT1, KIF7, KPTN, L1CAM, MED12, MLC1, MTOR, NFIX, NSD1, OFD1, PDGFRB, PIK3CA, PIK3R2, PTEN, RAB39B, RIN2, RNF125, RNF135, SETD2, SHANK3, STRADA, SUZ12, TBC1D7, ZNF469, HIST1H1E, ASPA, KIDINS220, MSL3, BMP4, BRAF, DEPDC5, FIBP, FOXP1, HRAS, KRAS, MAP2K1, MAP2K2, MYCN, NFIA, NFIB, NOTCH2NL, NRAS, ODC1, PTCH1, PTPN11, RAF1, RHEB, ROR2, SHOC2, SOS1, SPRED1, SUFU, SZT2, TSC1, TSC2, ZBTB20); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.06.2019

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Primäre Mikrozephalie (MCPH) (Panel MIC) (ASPM, CDK5RAP2, CEP152, MCPH1, ANKLE2, ARX, BRAT1, CASC5, CASK, CDC6, CDK5, CDK6, CDT1, CENPE, CENPJ, CEP135, CEP63, CKAP2L, DCX, DDX11, DHCR7, DNA2, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MFSD2A, NBN, NHEJ1, NIN, ORC1, ORC4, ORC6, PAFAH1B1 (LIS1), PCNA, PCNT, PHC1, PPM1D/WIP1, RAD50, RBBP8, RELN, RMI1, RNU4atac, RTTN, SASS6, STIL, TRAIP, TUBA1A, WDR62, XRCC4, XRCC9, ZNF335, EFTUD2, OSGEP, TP53RK, TPRKB, LAGE3, PNKP, WDR73, NCAPH2, CIT, NSMCE2, NEK3, CEP350, FANCM, CSNK2B, PYCR2, WRN, NUP133, NUP107, WDR4, ZEB2, TRMT1, TRAPPC9, NMT3A, DYNC1I2, VPS51, NDE1, BUB1B, KIF5C, KIF2A, TUBG1, TUBB2B, TBCD, POC1A , CHAMP1, MRE11A, STAMBP, COPB2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Seckel-Syndrom (SKS) (Panel MIC) (CEP63, CEP152, CENPJ, CDK5RAP2, DNA2, RBBP8, TRAIP, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK6, CDT1, CENPE, CEP135, CKAP2L, DCX, DDX11, DHCR7, ERCC4, ERCC6, FAAP95 (FANCB), FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NEHJ1, NBN, NIN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RELN, RMI1, RNU4atac, RTTN, SASS6, SLX4, STIL, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020

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LIG4-Syndrom (Panel MIC) (LIG4, XRCC4, BLM, DDX11, FANCD2, NBN, NHEJ1, PCNA, PPM1D, RAD50, ANKLE2, ARX, ASPM, ATR, ATRIP, B9D1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, MCPH1, MFSD2A, NIN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNT, PHC1, PHF6, PHF9, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC9, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Meckel-Syndrom (MKS) (Panel MIC) (CC2D2A, CEP290, MKS1, NPHP3, RPGRIP1L, TMEM216, B9D1, B9D2, KIF14, TCTN2, TMEM67, TMEM231); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Kleinwuchs, mikrozephaler osteodysplastischer primordialer, Typ I und Typ 2 (MOPD1 und MOPD2 (MOPD)) (Panel MIC) (PCNT, RNU4ATAC, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RMI1, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020

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Meier-Gorlin-Syndrom (MGS) (Panel MIC) (CDT1, CDC6, GMNN, ORC1, ORC4, ORC6, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDK5, CDK5RAP2, CDK6, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RNU4ATAC RMI1, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Nijmegen-Breakage-Syndrom (NBS) (Panel MIC) (NBN, RAD50, DDX11, BLM, FANCD2, LIG4, NHEJ1, PCNA, PPM1D, XRCC4, ANKLE2, ARX, ASPM, ATR, ATRIP, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, MCPH1, MFSD2A, NIN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNT, PHC1, PHF6, PHF9, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC9, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Fanconi-Anämie (FA) (Panel MIC) (BRCA2, FANCA, FANCC, FANCD2, FANCE, XRCC9 (FANCG), ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95 (FANCB), FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PCNT, PALB2, PCNA, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020

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37073 Göttingen

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Bloom-Syndrom (BS) (Panel MIC) (BLM, DDX11, FANCA, FANCC, NBN, RAD50, RMI1, XRCC9 (FANCG), ANKLE2, ARX, ASPM, ATR, ATRIP, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCD2, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD51C, RBBP8, RELN, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Zerebro-Okulo-Fazio-Skeletales-Syndrom (COFS) (Panel MIC) (ERCC1, ERCC2, ERCC5, ERCC6, ERCC8, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CDK5RAP2, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DNA2, ERCC4, FAAP95, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, GMNN, KMT2B, LIG4, NHEJ1, NBN, ORC1, ORC4, ORC6, PALB2, PIEZO2, PCNA, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RMI1, RTTN, SLX4, STIL, TRAIP, WDR62, XRCC4, XRCC9); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Filippi-Syndrom (FS) (Panel MIC) (CKAP2L, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, DDX11, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020
Galloway-Mowat-Syndrom (GMS) (Panel MIC) (LAGE3, OSGEP, TP53RK, TPRKB); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		22.12.2020

Hutchinson-Gilford-Progerie-Syndrom (HGPS) (Panel PROG) (LMNA, BANF1, FBN1, NARF, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1 ,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LIPE, LMNB1, LTPB4, MRE11A, NAA10, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, WRN, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Mandibuloakrale Dysplasie (MAD) (Panel PROG) (LMNA, POLD1, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, WRN, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

Nestor-Guillermo-Progerie-Syndrom (NGPS) (Panel PROG) (BANF1, LMNA, FBN1, NARF, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LIPE, LMNB1,LTPB4, MRE11A, NAA10, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Wiedemann-Rautenstrauch-Syndrom (WRS) (Panel PROG) (ANO6, CAV1, FBN1, LMNA, BANF1, PYCR1, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ATM, ATP6V0A2, ATP7A, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LIPE, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

Werner-Syndrom (WS) (Panel PROG) (WRN, LMNA, POLD1, SPRTN, ZMPST24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1, BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Ruijs-Aalfs-Syndrom (RAS) (Panel PROG) (SPRTN, WRN, LMNA, POLD1, ZMPST24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

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37073 Göttingen

Untersuchungsverfahren

Dokument ID: 145229
Stand: 31.10.2025

Cockayne-Syndrom (CS) (Panel PROG) (ERCC6, ERCC8, ERCC4, ERCC5, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Xeroderma pigmentosum (XP) (Panel PROG) (DDB2, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DKC1, EFEMP2, ELN, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

Bloom-Syndrom mit progeroiden Symptomen (BSPS) (Panel PROG) (BLM, PRKDC, SPRTN, WRN, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Lipodystrophie (LD) (Panel PROG) (AGPAT2, BSCL2, CAV1, CIDEC, LIPE, PLIN1, PPARG, PTRF, FBN1, LMNA, ZMPSTE24, ACD, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1, CHD6, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, POLD1, POLH, PRKDC, PTDSS1, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

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Untersuchungsverfahren

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Marfan-Lipodystrophie-Syndrom (MLS) (Panel PROG) (FBN1, AGPAT2, B3GALT6, B4GALT7, BSCL2, CAV1, LIPE, LMNA, PLIN1, PIK3R1, PPARG, PTRF, ACD, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, BLM, BRCA2, BRIP1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, POLD1, POLH, PRKDC, PTDSS1, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Short-Syndrom (STS) (Panel PROG) (PIK3R1, AGPAT2, BSCL2, CAV1, CIDEC, FBN1, LIPE, LMNA, PLIN1, PPARG, PTRF, ACD, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1, CHD6, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, POLD1, POLH, PRKDC, PTDSS1, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

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Keppen-Lubinsky-Syndrom (KLS) (Panel PROG) (KCNJ6, ANO6, CAV1, LMNA, PIK3R1, POLD1, PYCR1, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, LIPE, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Progerie Typ Penttinen (PTP) (Panel PROG) (PDGFRB, B3GALT6, B4GALT7, CAV1, FBN1, LIPE, LMNA, PLIN1, PIK3R1, PPARG, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, BLM, BRCA2, BRIP1, BSCL2, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PEX11B, POLD1, POLH, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

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Dokument ID: 145229
Stand: 31.10.2025

Cutis laxa assoziierte Syndrome (CLS) (Panel PROG) (ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, LTBP4, PEX11B, PYCR1, ACD, AGPAT2, ANO6, ATM, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, HELLS, KCNJ6, LIPE, LMNA, LMNB1, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Lenz-Majewski-Syndrom (LMS) (Panel PROG) (PTDSS1, ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, LTBP4, PEX11B, PYCR1, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPATG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

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Rothmund-Thomson-Syndrom (RTS) (Panel PROG) (RECQL4, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Dyskeratosis Congenita (DKC) (Panel PROG) (ACD, DKC1, NOLA2, NOLA3, PARN, RTEL1, TERC, TERT, TINF2, WRAP53, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, PALB2, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, SLX4, SMC2, SMC4, SPRTN, TINF2, TFAP2A, UBE2T, WFS1, WRN, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019

Ataxia-Teleangiektasia/Louis-Bar-Syndrom (LBS) (Panel PROG) (ATM, MRE11A, PCNA, ACD, AGPAT2, ALDH18A1, ANO6, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1,LTPB4, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		13.02.2019
Hereditäre Motorisch-Sensible Neuropathien/Charcot-Marie-Tooth-Neuropathien (HMSN/CMT), Distale Hereditäre Motorische Neuropathie (dHMN), Hereditäre Sensible und Autonome Neuropathien (HSAN) (Panel HMSN) (AARS, AIFM1, ARHGEF10, ATL1, ATL3, ATP7A, BSCL2, CCT5, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, 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	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		14.04.2021
Hereditäre Neuropathie mit Neigung zu Druckparesen (HNPP) (Panel HMSN) (PMP22); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		14.04.2021

Kabuki-Syndrom (Panel SYN) (KMT2D, KDM6A, RAP1A, RAP1B); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Rasopathien/Noonan-Syndrom (Panel SYN) (PTPN11, A2ML1, BRAF, CBL, CDC42, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS,RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, SPRED1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Costello-Syndrom (CSTLO) (Panel SYN) (HRAS); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Leopard-Syndrom (LPRD) (Panel SYN) (BRAF, PTPN11,RAF1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Kardiofaziokutanes Syndrom (CFC) (Panel SYN) (BRAF, MAP2K1, MAP2K2, KRAS); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
CHARGE-Syndrom (Panel SYN) (CHD7, SEMA3E, TBX1, TBX22); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
DiGeorge-Syndrom (DGS) (Panel SYN) (TBX1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Abruzzo-Erickson-Syndrom (ABERS), CHARGE-like Syndrom (Panel SYN) (TBX22); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Neurofibromatose (Panel SYN) (NF1, NF2, SPRED1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Legius-Syndrom (Panel SYN) (SPRED1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Robinow-Syndrom (Panel SYN) (WNT5A, DVL1, DVL3, ROR2, FGD1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Aarskog-Syndrom (Panel SYN) (FGD1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Cornelia-de-Lange-Syndrom (Panel SYN) (NIPBL, DXS423E / SMC1A, SMC3, RAD21, HDAC8, AFF4, SETD5, KMT2A, MAU2, BRD4); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020

CHOPS-Syndrom (Panel SYN) (AFF4); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
KBG-Syndrom (KBGS) (Panel SYN) (ANKRD11); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Trichorhinophalangeales Syndrom 1 und 3 (TRPS1 und TRPS3) (Panel SYN) (TRPS1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Coffin-Siris-Syndrom (Panel SYN) (ARID1B, ARID1A, ARID2, DPF2, SMARCB1, SMARCA4, SMARCE1, SMARCA2, SOX11); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Nicolaides-Baraitser-Syndrom (NCBRS) (Panel SYN) (SMARCA2); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Baraitser-Winter-Syndrom (BRWS1) (Panel SYN) (ACTB, ACTG1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Borjeson-Forssman-Lehmann-Syndrom (BFLS) (Panel SYN) (PHF6); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Cutis laxa (ADCL1) / Williams-Beuren-Syndrom (WBS) (Panel SYN) (ELN); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Simpson-Golabi-Behmel-Syndrom (SGBS) (Panel SYN) (GPC3, GPC4, OFD1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Townes-Brocks-Syndrom (TBS) (Panel SYN) (SALL1, DACT1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Duane-Radial Ray-Syndrom (DRRS) / Okihiro-Syndrom (Panel SYN) (SALL4); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Branchiooto(renales) Syndrom 1 und 2 (BOR1, BOR2) (Panel SYN) (EYA1, SIX1, SIX5); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Rubinstein-Taybi-Syndrom (RSTS) (Panel SYN) (CREBBP, EP300); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Floating-Harbor-Syndrom (FLHS) (Panel SYN) (SRCAP); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Coffin-Lowry-Syndrom (CLS) (Panel SYN) (RPS6KA3); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Okulodentodigitale Dysplasie (ODDD) (Panel SYN) (GJA1)	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020

Ritschner-Schintzel-Syndrom (RTSC) (Panel SYN) (WASHC5/KIAA0196, CCDC22); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Lujan-Fryns-Syndrom (LFS), Opitz-Kaveggia-Syndrom (OKS) (Panel SYN) (MED12); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Cohen-Syndrom (COH) (Panel SYN) (VPS13B); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Mental retardation, autosomal dominant (MRD32) (Panel SYN) (KAT6A); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Glass-Syndrom (Panel SYN) (SATB2); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Schuurs-Hoeijmakers-Syndrom (SHMS) (Panel SYN) (PACS1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Odgen-Syndrom, Lenz-Microphthalmie-Syndrom (Panel SYN) (NAA10); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		13.05.2020
Kraniosynostosen (Panel CRAN) (FGFR1, FGFR2, FGFR3, GLI3, MEGF8, RAB23, TWIST1, ADAMTS10, ALX1, ALX3, ALX4, CD96, EFNB1, ERF, IFT122, IFT43, IL11RA, LRP2, MN1, P4HB, POR, RECQL4, RUNX2, SEC24D, SKI, SMO, WDR19, WDR35, ZIC1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIumina, Nextseq550; Illumina, NovaSeq6000		x		23.11.2016
Kardiomyopathie, dilatativ (DCM) (Panel CARDIO) (DMD, LMNA, MYBPC3, MYH7, TNNT2, TPM1, ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CASQ2, CRYAB, CSRP3, CFT1, DES, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKTN, GATAD1, ILK, LAMA2, LAMA4, LAMP2, LDB3, MURC, MYH6, MYL2, MYL3, MYPN, NEXN, NPPA, PDLIM3, PLN, PRDM16, RAF1, RBM20, SCN5A, SGCD, TAZ, TBX20, TBX5, TCAP, TMPO, TNNC1, TNNI3, TTR, VCL); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Kardiomyopathie, hypertroph (HCM) (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	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019

Arrythmogene rechtsventrikuläre Kardiomyopathie / Dysplasie (ARVC/D) (Panel CARDIO) (DSC2, DSG2, DSP, JUP, PKP2, TGFB3, TMEM43, CASQ2, RYR2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Non-compaction Kardiomyopathie (NCCM) (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Kardiomyopathie, restriktiv (RCM) (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Morbus Fabry (Panel CARDIO) (GLA); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Catecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) (Panel CARDIO) (CASQ2, CALM1, KCNE1, KCNJ2, RYR2, TRDN); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Long QT-Syndrom (LQT) (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Short QT-Syndrom (SQT) (Panel CARDIO) (CACNA1C, KCNH2, KCNQ1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Brugada-Syndrom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Hypercholestrinämie (LDLR_Defizienz) (Panel CARDIO) (LDLR); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Hypercholesterinämie (primär) (Panel CARDIO) (APOB, PCSK9, LDLRAP1); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Amyloidose (erbliche Form) (Panel CARDIO) (TTR); SNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019

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Untersuchungsverfahren

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Marfan-Syndrom (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-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	Illumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Ehlers-Danlos-Syndrom (vaskuläre Form) (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Thorakale Aortenerweiterung (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Bindegewebserkrankung mit Gefäßbeteiligung (Panel CARDIO) (ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, LTPB2, MYH11, MYLK, MYLK2, NOTCH1, SLC2A10, SMAD3, SMAD4, TGFB2, TGFBR1, TGFBR2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		15.04.2019
Maturity-onset diabetes of the young (MODY) (Panel UNI) (ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
Familiärer Hyperinsulinismus / Hyperinsulinämische Hypoglykämie (HHF) (Panel UNI) (ABCC8, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, SLC16A1, UCP2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
Familiäre Hypercholesterinämie (FH) (Panel UNI) (APOB, LDLR, LDLRAP1, PCSK9); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
Familiäre hypokalzurische Hyperkalzämie (HHC) (Panel UNI) (CASR, AP2S1, GNA11); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
Kallmann-Syndrom / Isolierter Hypogonadotroper Hypogonadismus (IHH) (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
Morbus Osler / Hereditäre Hämorrhagische Teleangiektasie (HHT) (Panel UNI) (ACVRL1, ENG, SMAD4, GDF2, RASA1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021

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Cystische Fibrose (CF) (Panel UNI) CFTR	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
Retinitis pigmentosa, autosomal-dominant und x-gekoppelt (Panel AUGE) (ARL3, BEST1, CA4, CACNA1F, CRX, FSCN2, GUCA1B, HK1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, OFD1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, SPP2, TOPORS); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Retinitis pigmentosa, autosomal-rezessiv und x-gekoppelt (Panel AUGE) (ABCA4, ADGRA3, ADIPOR1, AGBL5, AHI1, AIPL1, ARHGEF18, ARL2BP, ARL6, BBS2, C8ORF37, CDHR1, CEP290, CERKL, CLCC1, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CWC27, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, GNAT1, GNPTG, GUCY2D, HGSNAT, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIF11, KIZ, LRAT, MAK, MERTK, MFRP, NEK2, NEUROD1, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF31, RBP3, RCBTB1, RDH11, RDH12, REEP6, RGR, RHO, RLBP1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, SAG, SAMD11, SLC7A14, SPATA7, TRNT1, TTC8, TUB, TULP1, USH2A, WDR19, ZNF408, ZNF513); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
ROSAH-Syndrom (Panel AUGE) (ALPK1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Mikrocornea, myopische chorioretinale Atrophy, Telecanthus (Panel AUGE) ADAMTS18	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Usher-Syndrom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023

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Refsum-Syndrom / Zellweger-Syndrom-Spektrum (Refsum / Zellweger / Neonatale Adrenoleukodystrophie) (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Bardet-Biedl-Syndrom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Alagille-Syndrom (Panel AUGE) (JAG1, NOTCH2, BMP2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Leber'sche kongenitale Amaurose (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Kongenitale stationäre Nachtblindheit (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Zapfendystrophie (Zapfen-Stäbchen-Dystrophie) (Panel AUGE) (ABCA4, ADAM9, AIPL1, ALMS1, ATF6, BEST1, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP78, CEP250, CERKL, CFAP410, CNGA3, CNGB3, CNNM4, CRB1, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, NMNAT1, NR2E3, NRL, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RDH12, RGS9, RGS9BP, RIMS1, RPGR (inkl. ORF15), RPGRIP1, SEMA4A, TTLL5, TULP1, UNC119); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023

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Makuladystrophien / Morbus Stargardt (Panel AUGE) (ABCA4, BEST1, C1QTNF5, CDH3, CFH, CLN3, CNGB3, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, GUCA1A, GUCY2D, IMPG1, IMPG2, IRX1, MAPKAP3, MFSD8, PPT1, PRDM13, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTLL5); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Optikusatrophie und Leber´sche hereditäre Optikusneuropathie (LHON) (Panel AUGE) (MT-ND1, MT-ND4, MT-ND4L, MT-ND6, ACO2, AFG3L2, ANTXR1, ATP1A3, C12ORF65, CISD2, DNM1L, FDXR, MFN2, MTPAP, NBAS, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, WFS1, YME1L1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Flecked Retina Erkrankungen (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Vitreoretinopathien (Familiäre Exsudative Vitreoretinopathie) (Panel AUGE) (ATOH7, BEST1, CAPN5, COL18A1, COL2A1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RCBTB1, TSPAN12, VCAN, ZNF408); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Senior-Loken-Syndrom (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Achromatopsie (Panel AUGE) (ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Myopie und Differentialdiagnosen (Panel AUGE) (COL11A1, COL11A2, COL18A1, COL2A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, FBN1, FZD4, LRP2, LRP5, P3H2, VCAN); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023

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Katarakt (Panel AUGE) (ABCB6, ABHD12, ADAMTSL4, AGK, BFSP1, BFSP2, CHMP4B, CLPB, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP51A1, CYP27A1, DNMBP, EPG5, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, GALK1, GALT, GCNT2, GEMIN4, GJA3, GJA8, GLA, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, LTBP2, MAF, MIP, MIR184, MYH9, NDP, NHS, OCRL, OPA3, P3H2, PAX6, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RECQL2, RECQL4, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TMEM114, UNC45B, VIM, VSX2, WFS1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Mikrophthalmie-Anophthalmie-Kolobom Komplex (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	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Linsenluxation (Panel AUGE) (ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, CBS, COL11A1, COL18A1, COL2A1, FBN1, LTBP2, P3H2, VCAN, VSX2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Hornhautdystrophien/Keratoconus (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	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Septo-optische Dysplasie (Panel AUGE) (FGFR1, HESX1, OTX2, PROKR2, SOX2, SOX3); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023

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Kongenitales Glaukom / Axenfeld-Rieger-Syndrom (Panel AUGE) (ASB10, COL4A1, CYP1B1, DDX58, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, SBF2, SH3PXD2B, TBK1, TEK, WDR36); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Vorderkammerdysgenesien / Axenfeld-Rieger-Syndrom (Panel AUGE) (ASPH, B3GLCT, COL4A1, COL4A2, CYP1B1, EYA1, FOXC1, FOXE3, GPATCH3, HMX1, LTBP2, MYOC, PAX6, PITX2, PXDN, SLC38A8); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Albinismus (syndromal) (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	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		06.11.2023
Osteoporose/Erniedrigte Knochenmineraldichte (OEN) (Panel SKELETT) (ALPL, COL1A1, COL1A2, DKK1, IFITM5, LRP5, LRP6, MBTPS2, PLS3, SGMS2, WNT1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Osteogenesis Imperfecta (OI) / frühmanifeste Frakturneigung (z.B. Hypophosphatasie, Osteoporose-Pseudogliom) (Panel SKELETT) (ALPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, GORAB, IFITM5, KDELR2, LRP5, MESD, P3H1, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TENT5A, TMEM38B, WNT1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Bindegewebserkrankungen mit Kontrakturen (z.B. Bruck-Syndrom, Weill-Marchesani-Syndrom) (Panel SKELETT) (ADAMTS10, FBN1, FBN2, FKBP10, PLOD2, PLOD3); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Ehlers-Danlos-Syndrom (EDS) (Panel SKELETT) (B3GALT6, B4GALT7, CHST14; COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, PLOD1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Störungen des Kalzium- und Phosphatstoffwechsels (z.B. hypophosphatämische Rachitis, Pseudohypoparathyreoidismus) (Panel SKELETT) (CASR, CLCN5, DMP1, ENPP1, FGF23, GALNT3, GNAS, KL, PHEX, SAMD9, SLC9A3R1, SLC34A1, SLC34A3, STX16); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022

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Osteopetrose / erhöhte Knochenmineraldichte (z.B. Dysosteosklerose, Osteopoikilose, Raine-Syndrom, Sklerosteose) (Panel SKELETT) (AMER1, ANKH, CA2, CLCN7, CTSK, FAM20C, LEMD3, LRP5, LRRK1, OSTM1, PLEKHM1, SNX10, SOST, TCIRG1, TGFB1, TNFSF11, TNFRSF11A); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Multiple epiphysäre Dysplasie / Früharthrose (z.B. pseudorheumatoide Dysplasie, Pachydermoperiostosis) (Panel SKELETT) (ACAN, CANT1, CCN6 (WISP3), COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2, SLCO2A1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Chondrodysplasien mit ausgeprägtem Kleinwuchs (z.B. Achondroplasie, Rubinstein-Taybi-Syndrom, kampomele Dysplasie) (Panel SKELETT) (ACAN, CREBBP, CUL7, EP300, FGFR3, PTHR1, RMRP, RNU4ATAC, SHOX, SLC26A2, SMARCAL1, SOX9, TRIP11); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Stickler-Syndrom und Sonstige Chondrodysplasien (z.B. kleidokraniale Dysplasie, trichorhinophalangeales Syndrom) (Panel SKELETT) (COL2A1, COL9A1, COL9A2, COL10A1, COL11A1, COL11A2, RUNX2, TRAPPC2, TRPS1, TRPS2, TRPV4); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Dysostosen der Extremitäten (z.B. Cenani-Lenz-Syndrom, Holt-Oram-Syndrom, Okihiro-Syndrom, Polydaktylie) (Panel SKELETT) (ESCO, GLI3, LRP4, SALL1, SALL4, TBX3, TBX5, WNT3, ZRS-Region); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Kraniofaziale Dysostosen (z.B. frontonasale Dysplasie, parietale Foramina, Treacher-Collins-Syndrom) (Panel SKELETT) (ALX1, ALX3, ALX4, BMP4, DHOD, EFNB1, EFTUD2, GLI3, MN1, MSX2, POLR1C, POLR1D, SF3B4, TCOF, ZSWIM6); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Kraniosynostose (z.B. Apert-Syndrom, Muenke-Syndrom, Pfeiffer-Syndrom, Seathre-Chotzen-Syndrom) (Panel SKELETT) (ERF, FGFR1, FGFR2, FGFR3, IL11RA, P4HB, RECQL4, SEC24D, SKI, TCF12, TWIST1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022

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Kraniosynostose mit prominenter extraskelettaler Symptomatik (z.B. kranioektodermale Dysplasien, Carpenter-Syndrom) (Panel SKELETT) (CD96, EVC, EVC2, IFT22, IFT43, LRP2, MEGF8, POR, RAB23, RECQL4, WDR35, ZIC1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		11.07.2022
Ataxie, autosomal-dominant (Panel NEUROD) (AFG3L2, EEF2, FGF14, KCNC3, KCND3, PDYN, PUM1, PRKCG, SPTBN2, TGM6, TMEM240, TTBK2, VAMP1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Ataxie, autosomal-rezessiv (Panel NEUROD) (ANO10, APTX, PHYH, PITRM1, PNPLA6, POLG, SETX, SPG7, STUB1, TTPA); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Ataxie, unbestimmt (Panel NEUROD) (ANO10, APTX, CYP27A1, FGF14, PHYH, PRKCG, SETX, SPG7, TTBK2, TTPA); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Early-onset Ataxie (Panel NEUROD) (APTX, FXN, POU4F1, SACS); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Episodische Ataxie (Panel NEUROD) (CACNA1A, CACNB4, KCNA1,SCN2A, SLC1A3); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Parkinson-Erkrankung, autosomal-dominant (Panel NEUROD) (CHCHD2, GBA, LRRK2, TMEM230, SNCA, VPS35); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Parkinson-Erkrankung, autosomal-rezessiv (Panel NEUROD) (ATP13A2, DNAJC6, FBXO7, PARK7, PLA2G6, PINK1, PRKN, SLC30A10, SLC6A3, SYNJ1, VPS13C); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Parkinson-Erkrankung, unbestimmt (Panel NEUROD) (ATP13A2, CHCHD2, FBXO7, GCH1, LRRK2, PRKN, PARK7, PINK1, PLA2G6, SNCA, UCHL1, TH); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Atypische Parkinson-Erkrankung (Panel NEUROD) (ATP13A2, ATP1A3, DCTN1, DNAJC6, FBXO7, GCH1, LRP10, MAPT, PLA2G6, SLC30A10, SYNJ1, TH, VPS13C); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023

Parkinson-Dystonie (Panel NEUROD) (ATP1A3, DNAJC12, GCH1, PLA2G6, PRKRA, SLC30A10, SPR, SLC39A14, SLC6A3, TAF1, TH); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Dystonie (Basis) (Panel NEUROD) (ATP1A3, GCH1, GNAL, ECHS1, KMT2B, PNKD, PARK2, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TH, THAP1, TOR1A); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Primäre Torsionsdystonie (Panel NEUROD) (ANO3, CIZ1, COL6A3, GNAL, HPCA, THAP1, TOR1A, TUBB4A); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Dystonie-Plus-Syndrom (Panel NEUROD) (ATP1A3, BCAP31, COX20, FTL, GCH1, KIF1C, PRKRA, SLC30A10, SGCE, SPR, TAF1, TH, TUBB4A); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Choreatiforme Bewegungsstörungen (Dyskinesie) (Panel NEUROD) (ADCY5, KCNA1, KCNMA1, NKX2-1, PDE10A, PDE8B, PNKD, PRNP, PRRT2, RNF216, SLC2A1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Neuronale Ceroidlipofuszinose (Panel NEUROD) (ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Neuroakanthozytose (Panel NEUROD) (PANK2, VPS13A, XK); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Morbus-Wilson (Panel NEUROD) (ATP7B); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Demenz (Basis) (Panel NEUROD) (APOE, APP, CHMP2B, CSF1R, GRN, ITM2B, MAPT, NOTCH3, PSEN1, SQSTM1, TARDBP, VCP); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Frontotemporale Demenz (FTD) (Panel NEUROD) (CHCHD10, CHMP2B, CSF1R, FUS, GRN, HNRNPA2B1, ITM2B, MAPT, PSEN1, PSEN2, SQSTM1, TARDBP, TBK1, TBP, TREM2, UBQLN2, VCP); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Alzheimer-Erkrankung (Panel NEUROD) (APOE, APP, PSEN1, PSEN2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023

Vaskulär-bedingte Demenz (Panel NEUROD) (COL4A1, HTRA1, NOTCH3, TREM2, TREX1, TYROBP); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Amyotrophe Lateralsklerose (ALS) (Panel NEUROD) (ALS2, ANG, CHCHD10, CHMP2B, FIG4, FUS, MATR3, OPTN, PFN1, SOD1, TARDBP, TUBA4A, UBQLN2, VAPB, VCP); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Hereditäre spastische Paraplegie (HSP), autosomal-dominant (Panel NEUROD) (ATL1 (SPG3A), BICD2, BSCL2, CPT1C, HSPD1, WASHC5 (SPG8), KIF5A, KPNA3, NIPA1, REEP1 (SPG31), RTN2, SLC33A1, SPAST* (SPG4), UBAP1, VAMP1, ZFYVE27); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Hereditäre spastische Paraplegie (HSP), autosomal-rezessiv (Panel NEUROD) (CYP7B1 (SPG5A), FA2H, GJC2, SPG11, SPG7, ZFYVE26); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Hereditäre spastische Paraplegie (HSP), X-chromosomal (Panel NEUROD) (ABCD1, L1CAM, PLP1, SLC16A2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Hereditäre spastische Paraplegie (HSP), unbestimmt (Basis) (Panel NEUROD) (ATL1 (SPG3A), CYP7B1 (SPG5A), SPG11, KIF1A, KIF5A, REEP1 (SPG31), SPAST (SPG4), SPG7); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023

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Untersuchungsverfahren

Hereditäre spastische Paraplegie (HSP), unbestimmt (gesamt) (Panel NEUROD) (ABCD1, ADAR, AFG3L2, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, B4GALNT1, BICD2, BSCL2, MTRFR, C19orf12, CCT5, CPT1C, CYP2U1, DDHD1, DDHD2, DNM2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAM134B, FARS2, FLRT1, FUS, GAD1, GBA2, GJC2, GRID2, HPDL, IBA57, IFIH1, KIDINS220, WASHC5 (SPG8), KIF1C, KLC2, KLC4, L1CAM, LYST, MAG, MARS2, NIPA1, NT5C2, PGAP1, PLP1*, PMCA4, PNPLA6, RAB3GAP2, REEP2, RNASEH2B, RTN2, SETX, SLC16A2, SLC33A1, SOD1, SOX10, SPATA5L1, SPG20, SPG21,TARDBP, TECPR2, TFG, TUBB4A, USP8, VAMP1, VCP, VPS37A, WDR48, ZFR, ZFYVE26, ZFYVE27); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Basalganglien-Kalzifizierungen (Panel NEUROD) (ADAR, CA2, COL4A1, CTC1, ERCC6, ERCC8, GALC, IFIH1, OCLN, PDGFRB, PDGFB, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC20A2, TREM2, TREX1, TYROBP, XPR1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Neurodegeneration mit Eisenablagerungen (NBIA) (Panel NEUROD) (ATP13A2, C19orf12, CCNF, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Infantile und juvenile Leukodystrophien (Panel NEUROD) (ABCD1, AIMP1, ARSA, ASPA, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, DARS2, GALC, GFAP, GJC2, HEPACAM, MLC1, PLP1, PSAP, RNASET2, TMEM63A, TUBB4A); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Adulte Leukodystrophien (Panel NEUROD) (ABCD1, ARSA, CSF1R, CYP27A1, DARS2, EIF2B5, GALC, GFAP, HTRA1, LMNB1, MLC1, NOTCH3); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023

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Untersuchungsverfahren

Leukodystrophien und Leukenzephalopathien (Panel NEUROD) (AARS1, AARS2, ACBD5, ACOX1, ADAR, ALDH3A2, BCAP31, CLCN2, CTC1, CTSA, EARS2, EIF2AK2, FAM126A, FUCA1, GBE1, GCDH, HIKESHI, HSD17B4, HSPD1, IFIH1, L2HGDH, NAXE, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PLEKHG2, POLR1C, POLR3A, POLR3B, PYCR2, RARS, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, SLC16A2, SLC17A5, SNORD118, SOX10, STN1, SUMF1,TMEM106B, TREX1, VPS11); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		24.05.2023
Ichtyosis (nicht syndromal/syndromal) (Panel HAUT) (FLG, STS, ABCA12, ALOX12B, ALOXE3, CDSN, CERS3, CHST8, CSTA, CYP4F22, FLG2, GJB3, GJB4, KRT1, KRT10, KRT2, LIPN, NIPAL4, PNPLA1, SDR9C7, SERPINB8, ST14, SULT2B1, TGM1, TGM5, ABHD5, ALDH3A2, AP1B1, AP1S1, CLDN1, EBP, ELOVL4, GJB2, MBTPS2, NSDHL, PHYH, PIGL, SLC27A4, SNAP29, SPINK5, ST14); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Ektodermale Dysplasien (Panel HAUT) (CDH3, CST6, DLX3, EDA, EDAR, EDARADD, GJA1, GJB6, HOXC13, IFT122, IFT43, IKBKG, KDF1, KREMEN1, KRT74, KRT85, MSX1, NECTIN1, NECTIN4, NFKBIA, PORCN, TP63, TSPEAR, WDR19, WDR35, WNT10A); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Epidermolysis bullosa (Panel HAUT) (CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSP, DST, EXPH5, FLG2, ITGA3, ITGA6, ITGB4, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1, PLEC1, SERPINB8, TGM5); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Tumorerkrankungen der Haut (Gorlin-Goltz-Syndrom, Malignes Melanom, Birt-Hogg-Dube-Syndrom, Familiäre Leiomyomatose mit Nirenzellkarzinom) (Panel HAUT) (PTCH1, PTCH2, SUFU, CDK4, CDKN2A, MC1R, MITF, POT1, TYR, FLCN, FH); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022

Bindegewebserkrankungen (Marfan-Syndrom, Cutis laxa, Pseudoxanthoma elasticum) (Panel HAUT) (CBS, FBN1, FBN2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, EFEMP2, ELN, FBLN5, LTBP4, PYCR1, ABCC6, XYLT1, XYLT2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Neurokutane Syndrome (Neurofibromatose, Tuberöse Sklerose) (Panel HAUT) (LZTR1, NF1, NF2, SMARCB1, SPRED1, TSC1, TSC2); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Acne inversa (Panel HAUT) (PSENEN, NCSTN, PSEN1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Pigmentierunsstörungen (Incontinentia pigmenti, oculokutaner Albinismus, syndromale Hypopigmentierungsstörungen, Morbus Dowling-Degos und andere Hyperpigmentierungsstörungen) (Panel HAUT) (IKBKG, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1, AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LYST, MITF, MLPH, MYO5A, PAX3, RAB27A, SNAI2, SOX10, ABCB6, ADAM10, ADAR, KITLG, KRT14, KRT5, POFUT1, POGLUT1, POLA1, PSENEN, SASH1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Hereditäres Angioödem (Panel HAUT) (C1NH, F12); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Hereditäres Lymphödem (Panel HAUT) (CCBE1, FAT4, FLT4, FOXC2, GJC2, PIEZO1, VEGFC); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Palmoplantare Keratosen (Panel HAUT) (AAGAB, AQP5, CAST, CTSB, CTSC, DSG1, DSP, FERMT1, GJB2, JUP, KANK2, KRT1, KRT10, KRT14, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT9, LOR, MBTPS2, PKP1, POMP, RHBDF2, RSPO1, SERPINB7, SLURP1, SMARCAD1, TAT, TRPV3); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIIIlumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022

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Follikuläre Verhornungsstörungen (Panel HAUT) (ATP2A2, ATP2C1, MBTPS2, RIPK4); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Trichothiodystrophie (Panel HAUT) (ERCC2, ERCC3, GTF2E2, GTF2H5, MPLKIP, RNF113A, TARS1); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Haaranomalien (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, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Nageldysplasien (Panel HAUT) (COL7A1, EDAR, FZD6, HOXC13, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT74, KRT85, PLCD1, RIPK4, RSPO4); SNV, CNV	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		19.10.2022
Erbliche Hörstörungen (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,	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021

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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	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline; MLPA	AA-112, V005; AA-113, V004; AA-156, V003; AA-157, V002; AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-502, V002; AA-504, V003; AA-506, V004; Varvis V1.25; ICA V2	ThermoFisher Scientific, SeqStudio; ThermoFisher Scientific, Genetic Analyzer 3500xlIllumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
Erblich bedingte mitochodriale Erkrankungen (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	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	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	Illumina, Nextseq550; Illumina, NovaSeq6000		x		21.04.2023
Whole Exome Sequencing (WES); SNV, CNV (Agilent SureSelect Human All Exon V8)	Blut, DNA; DNA	Sequence capture; Sequencing-by synthesis; custom pipeline;	AA-209, V003; AA-220, V002; AA-247, V004; AA-501, V002; AA-504, V003; AA-505, V003; AA-506, V004; AA-507, V003; Varvis V1.25; ICA V2	Illumina, Nextseq550; Illumina, NovaSeq6000		x		20.12.2021
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-220, V002; AA-247, V004; AA-501, V002; AA-504, V003; AA-506, V004; AA-508, V001; ICA V2	Illumina, NovaSeq6000		x		26.07.2024