

## Untersuchungsgebiet: Humangenetik

### Untersuchungsart:

#### Chromosomenanalyse

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version
Angeborener Chromosomensatz	Blut, Fruchtwasser, Chorionzotten oder andere Gewebeprobe <sup>a</sup>	Chromosomenbänderungsanalyse	va_Bearb_peripheres_Blut (07.04.2022) va_Bearb_Fruchtwasser (11.02.2022) va_Bearb_Chorionzotten (01.12.2023) va_Bearb_Fibroblasten (07.04.2022)
Angeborener Chromosomensatz	Blut, Fruchtwasser, Chorionzotten oder andere Gewebeprobe <sup>a</sup> , Mundschleimhautabstrich	Fluoreszenz-in-situ-Hybridisierung (FISH)	va_Bearb_peripheres_Blut (07.04.2022) va_Bearb_Fruchtwasser (11.02.2022) va_Bearb_Chorionzotten (01.12.2023) va_Bearb_Fibroblasten (07.04.2022) aa_FISH_Mundschleimhautabstrich (01.12.2023)
Erworbener Chromosomensatz	Blut, Knochenmark	Chromosomenbänderungsanalyse	va_Bearb_peripheres_Blut (07.04.2022) aa_Probenannahme_u_Ansatz_TG (01.12.2023) Praeparatherst_TG (23.10.2023) aa_Karyotypanalyse_TG (01.12.2023)
Erworbener Chromosomensatz	Blut, Knochenmark, Knochenmarkausstrich	Fluoreszenz-in-situ-Hybridisierung (FISH)	va_Bearb_peripheres_Blut (07.04.2022) va_Bearb_Fruchtwasser (11.02.2022) va_Bearb_Chorionzotten (01.12.2023) va_Bearb_Fibroblasten (07.04.2022) aa_FISH_Mundschleimhautabstrich (01.12.2023) aa_FISH_KM_Ausstrich (13.04.2022) aa_FISH_KM_u_Blut_aus_Kultur (13.04.2022)
Angeborener Chromosomensatz	Blut, DNA; DNA	molekulare Karyotypisierung mittels Array-CGH	va_Array_CGH (26.10.2022)

<sup>a</sup>Die Proben werden vom Labor ohne vorherige histologische Beurteilung bearbeitet und analysiert.

### Untersuchungsart:

#### Molekularbiologische Untersuchungen (Amplifikationsverfahren)\*\* [Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version
Angelman-Syndrom (15q11-q13 Region)	EDTA-Blut, DNA; DNA	MLPA (methylierungssensitiv)	va_MS_MLPA_PWAS (23.10.2022)
Prader-Willi-Syndrom (15q11-q13 Region)	EDTA-Blut, DNA; DNA	MLPA (methylierungssensitiv)	va_MS_MLPA_PWAS (23.10.2022)
Ausschluss/ Nachweis mütterlicher Zellen im pränatalen Untersuchungsmaterial bzw. Abortdiagnostik	EDTA-Blut, Fruchtwasser, Chorionzotten oder andere Gewebeprobe <sup>a</sup> , Zellkulturen (aus Fruchtwasser, Chorionzotten und anderen Gewebeprobe <sup>a</sup> ), DNA; DNA	PCR, Fragmentanalyse	va_Ablauf_muetterliche_Kontamination (22.10.2022)
pränataler Schnelltest (STR-basiert)	EDTA-Blut, Fruchtwasser, Chorionzotten oder andere Gewebeprobe <sup>a</sup> , Zellkulturen (aus Fruchtwasser, Chorionzotten und anderen Gewebeprobe <sup>a</sup> ), DNA; DNA	PCR, Fragmentanalyse	va_STR_Praenataler_Schnelltest (28.09.2022)
Uniparentale Disomien der Chromosomen 6, 7, 11, 13, 14, 15, 18, 21	EDTA-Blut, Fruchtwasser, Chorionzotten, Zellkulturen (aus Fruchtwasser, Chorionzotten), DNA; DNA	PCR, Fragmentanalyse	va_MK_UPD_Abort_CE (21.10.2022)
Cystische Fibrose: Screening der häufigsten Mutationen (CFTR)	EDTA-Blut, Fruchtwasser, Chorionzotten, Zellkulturen (aus Fruchtwasser, Chorionzotten), DNA; DNA	SSP-PCR, Fragmentanalyse (CFTR-Assay)	va_CFTR_DE (22.10.2022)
Azoospermie (Deletionen im AZF-Locus: AZFa, AZFb, AZFc)	EDTA-Blut, DNA; DNA	SSP-PCR, Fragmentanalyse	va_AZF_Diagnostik (21.10.2022)
Fragiles-X-Syndrom (FMR1)	EDTA-Blut, DNA; DNA	PCR, Triplet Repeat-primed PCR, Fragmentanalyse	va_FRAX.docx (19.5.2025)
Chorea Huntington (Bestimmung der CAG/CCG-Repeats im HTT-Gen)	EDTA-Blut, DNA; DNA	Fragmentanalyse	va_Chorea_Huntington (21.10.2022)

DPYD-Defizienz / 5-FU-Toxizität (DPYD: dbSNP rs3918290, rs55886062, rs67376798, rs56038477)	EDTA-Blut, DNA; DNA	Loop Amplification, Fluoreszenzmarkierte Hybridisierungs sonden (Real-time PCR)	va_LAMP_DPYD (15.10.2021)
Whole Exome (WES)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080 ukj_ih_va_WGS 26.5.2025
Whole Genome (WGS)	EDTA-Blut, DNA; DNA	NGS (PCR-free library, sequencing-by-synthesis, custom pipeline, SNV, indel, CNV) MLPA, Sanger-Sequenzierung	varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Agammaglobulinämie (BLNK, BTK, CD79A, CD79B, FNIP1, IGLL1, PIK3CD, PIK3R1, SLC39A7, SPI1, TCF3, TOP2B)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Amelogenesis imperfecta (isoliert) (AMBN, AMELX, ENAM, WDR72, ACP4, COL17A1, DLX3, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMA3, LAMB3, MMP20, ODAPH, RELT, SLC24A4, SP6)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Amelogenesis imperfecta (syndromal) (CNNM4, FAM20C, LTBP3, ORAI1, PEX1, PEX26, PEX6, ROGD1, SLC10A7, SLC13A5, STIM1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Andere angeborene Immunitätsstörungen im Zusammenhang mit Leukozyten (IL18BP, IRF4)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Andere angeborene Immunitätsstörungen im Zusammenhang mit nicht hämatopoetischen Geweben (APOL1, CLCN7, HMOX1, NBAS, NCSTN, OSTM1, PLEKHM1, PSEN1, PSENE1, RANBP2, RPSA, SNX10, TCIRG1, TNFRSF11A, TNFRSF44)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Anfälligkeit für EBV und lymphoproliferative Erkrankungen (CARMIL2, CD27, CD70, CTPS1, MAGT1, PRKCD, RASGRP1, SH2D1A, TET2, TNFRSF9, XIAP)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Anfälligkeit für mukokutane Candidiasis (IL17F, IL17RA, IL17RC, MAPK8, STAT1, TRAF3IP2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Anfälligkeit für mykobakterielle Erkrankungen (MSMD) (CYBB, IFNG, IFNGR1, IFNGR2, IL12B, IL12RB1, IL12RB2, IL23R, IRF8, ISG15, JAK1, RORC, SPPL2A, STAT1, TBX21, TYK2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Anfälligkeit für schwere Virusinfektion (FCGR3A, IFIH1, IFNAR1, IFNAR2, IRF7, IRF9, NOS2, POLR3A, POLR3C, POLR3F, STAT1, STAT2, TLR7, ZNF1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

<p>Angeborene Neutropenie (CEBPE, CLPB, CSF3R, CXCR2, DNAJC21, EFL1, ELANE, G6PC3, GFI1, HAX1, HYOU1, JAGN1, LAMTOR2, SBDS, SLC37A4, SMARCD2, SRP54, TAFAZZIN, USB1, VPS13B, VPS45, WAS)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Angioödem (SERPING1, ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Autoimmunität mit oder ohne Lymphoproliferation (AIRE, ITCH, JAK1, PDCD1, PEPD, SOCS1, TPP2)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Bindegewebserkrankungen                  Gesamtpanel (Marfan-, Ehlers-Danlos-, Loeys-Dietz-Syndrom, TAAD, Differentialdiagnosen) (ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADAMTSL4, AEBP1, ALDH18A1, ARIH1, ASPH, ATP6V0A2, ATP6V1A, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, DSE, EFEMP2, ELN, EMILIN1, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GORAB, HCN4, HEY2, IPO8, LOX, LTBP1, LTBP2, LTBP3, LTBP4, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PYCR1, RIN2, ROBO3, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TAB2, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, TNXB, ZNF450)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Brust- und Eierstockkrebs (ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, NBN, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Cowden-Syndrom / Cowden-Syndrom-ähnliche Symptomatik (PTEN, AKT1, PIK3CA, SDHB, SDHC, SDHD, SEC23B)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Cystische Fibrose (CFTR)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Defekte des Inflammasoms (MEFV, MVK, NLR4, NLRP1, NLRP12, NLRP3, RIPK1)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>

Defekte des oxidativen Burst (CYBA, CYBB, CYBC1, G6PD, NCF1, NCF2, NCF4)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Endometriumkarzinom (EPCAM, MLH1, MSH2, MSH6, PMS2, MUTYH, NTHL1, POLD1, POLE, PTEN, STK11, TP53)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Epidermodysplasia verruciformis (HPV) (CIB1, CXCR4, TMC6, TMC8)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Familiäres thorakales Aortenaneurysma und Aortendissektion (TAAD) (ACTA2, FBN1, FOXE3, LOX, SMAD3, TGFBFR1, TGFBFR2, ABL1, ADAMTSL4, ARIH1, ASPH, BGN, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, EMILIN1, FBLN5, FBN2, FKBP14, FLNA, HCN4, HEY2, IPO8, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD4, SMAD6,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Fanconi-Anämie (BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, BLM, BRCA1, ERCC4, FAAP100, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFW3, SLX4, TOP3A, UBE2T, XRCC3)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
FHL-Syndrome mit Hypopigmentierung (AP3B1, AP3D1, LYST, RAB27A)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
FHL-Syndrome ohne Hypopigmentierung (inkl. ALPS, Canale-Smith-Syndrom) (CASP10, CASP8, FAAP24, FADD, FAS, FASLG, PRF1, RHOG, SLC7A7, STX11, STXBP2, UNC13D)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Gastrointestinaler Stromatumor (GIST) (KIT, PDGFRA, NF1, SDHA, SDHAF2, SDHB, SDHC, SDHD)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Hämochromatose, Differenzialdiagnosen (HFE, BMP6, CP, FTH1, FTL, HAMP, HJV, SLC40A1, TF, TFR2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

<p>Hereditäre Neuropathien (HMSN/HNPP/CMT) (GDAP1, GJB1, HINT1, MFN2, MPZ, PMP22, SH3TC2, SORD, AARS1, ABCA1, AIFM1, ARHGEF10, AT1L1, AT1L3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CADM3, CHCHD10, CNTNAP1, COA7, DCTN1, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FXN, GAN, GARS1, GLA, GNB4, HADHA, HADHB, HARS1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KARS1, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MME, MORC2, MPV17, MTMR2, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PHYH, PLEKHG5, PMP2, PNKP, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN11A, SCN9A, SCO2, SEPTIN9, SETX, SGPL1, SIGMAR1, SLC12A6, SLC25A46, SLC5A7, SPG11, SPTLC1, SPTLC2, SURF1, SYT2, TFG, TRIM2, TRPV4, TTR, VCP, VWA1,</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Hereditäre Schmerzkrankungen (CIP, SFN, PE, PEPD, FEPS1-3, FD) / HSAN (KIF1A, RETREG1, SCN9A, WNK1, AT1L1, AT1L3, DNMT1, DST, ELP1, FLVCR1, GLA, NGF, NTRK1, PRDM12, RAB7A, SCN10A, SCN11A, SPTLC1, SPTLC2, TECPR2, TRPA1, TTR, ZFHX2)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Hereditäre Spastische Paraplegie (HSP, SPG) (AT1L1, CYP7B1, KIF1A, REEP1, SPAST, SPG11, SPG7, ABCD1, ABHD16A, ADAR, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATP13A2, ATP2B4, B4GALNT1, BICD2, BSCL2, BTBD, C19orf12, CAPN1, CFAP276, COQ7, CPT1C, CYP27A1, CYP2U1, DARS1, DDHD1, DDHD2, DNM2, ENTPD1, ERLIN1, ERLIN2, FA2H, FAR1, FARS2, GAD1, GALC, GBA2, GCH1, GJC2, GRID2, HACE1, HPDL, HSPD1, IBA57, KCNA2, KDM5C, KIDINS220, KIF1C, KIF5A, KLC2, KLC4, L1CAM, MAG, MARS1, MMACHC, MTHFR, MTRFR, NEFL, NIPA1, NKX6-2, NT5C2, OPA3, PAH, PCYT2, PGAP1, PLP1, PNPLA6, POLR3A, PSEN1, REEP2, RNASEH2B, RNF170, RTN2, SACS, SELENOI, SERAC1, SLC16A2, SLC1A4, SLC2A1, SPART, SPG21, SPTAN1, TECPR2, TFG, TUBB4A, UBAP1, USP8, VCP, VPS13D, WASHC5, WDR45B, WDR48, ZFYVE26, ZFYVE27)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Hermansky-Pudlak-Syndrom (DD Chediak-Higashi-Syndrom, Griscelli-Syndrom) (AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, RAB27A)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Herpes-simplex-Enzephalitis (HSE) (ATG4A, DBR1, IRF3, MAP1LC3B2, SNORA31, TBK1, TICAM1, TLR3, TRAF3, UNC93B1)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022)                  va_NGS_IDT (07.03.2025)                  va_MLPA (23.10.2022)                  va_digitale_MLPA (21.06.2022)                  varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>

Herzrhythmusstörung— Erregungsleitungsstörungen / Sick- Sinus Syndrom / Brugada Syndrom (CCD, PCCD, SSS, SND, BRU) (CACNA1D, CASQ2, DES, DMD, EMD, GJC1, GLA, GNB2, GNB3, HCN4, KCNJ3, KCNJ5, KCNQ1, LAMP2, LMNA, MYH6, MYL4, NKX2-5, PRKAG2, RYR2, SCN5A, SGO1, TBX5, TNNI3K, TRPM4, TTR)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Herzrhythmusstörung - Katecholaminerge Polymorphe Ventrikuläre Tachykardie (CPVT) (CASQ2, RYR2, CALM1, CALM2, CALM3, KCNJ2, TECRL, TRDN)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Herzrhythmusstörung - Long-QT- Syndrom (LQTS) (KCNH2, KCNQ1, SCN5A, CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNE2, KCNJ2, TRDN)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Herzrhythmusstörung - Short-QT- Syndrom (SQTS) (KCNH2, KCNJ2, KCNQ1, SLC4A3)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Hyper-IgM-Syndrom (AICDA, CTNBL1, INO80, MSH6, TNFSF13, UNG, CD40, CD40LG, NFKBIA, PIK3CD, PIK3R1, PMS2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Immundysregulation mit Kolitis (ELF4, IL10, IL10RA, IL10RB, NFAT5, RIPK1, TGFB1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Isotyp-, Leichtketten- oder Funktionsdefizite mit normaler Anzahl von B-Zellen (CARD11)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kardiomyopathie—Dilatative- Kardiomyopathie (DCM)/ Arrhythmogene Kardiomyopathie- (ACM)/ Arrhythmogene- Rechtsventrikuläre Kardiomyopathie- (ARVC) (BAG3, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MYBPC3, MYH6, MYH7, PKP2, RBM20, SCN5A, TNNC1, TNNT2, TTN, ABCC9, ACTC1, ACTN2, CDH2, CSRP3, CTNNA3, DES, DMD, DOLK, EMD, EPG5, EYA4, FHL1, HAMP, HFE, HJV, IDH2, JPH2, LAMP2, NEXN, NKX2- 5, PLN, PPP1R13L, RYR2, SGCD, SLC40A1, SPEG, TAFAZZIN, TCAP, TFR2, TGFB3, TMEM43, TNNI3, TNNI3K, TPM1, VCL)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing- by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

Kardiomyopathie - Hypertrophe Kardiomyopathie (HCM) (ACTC1, ALPK3, FHOD3, MYBPC3, MYH7, MYL2, MYL3, PLN, TNNI3, TNNT2, TPM1, ABCC9, ACTN2, BAG3, BRAF, CACNA1C, CAV3, COX15, CRYAB, CSRP3, DES, FHL1, FLNC, FXN, GAA, GLA, HRAS, JPH2, KLHL24, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MYO6, NRAS, PRKAG2, PTPN11, RAF1, RASA2, RIT1, RRAS, SLC25A4, SOS1, SOS2, TNNC1, TRIM63, TTR)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kardiomyopathie - Non-Compaction-Kardiomyopathie (NCCM)/Linksventrikuläre Noncompaction-Kardiomyopathie (LVNC) (ACTC1, HCN4, MYBPC3, MYH7, PRDM16, RYR2, TNNT2, TTN, ACTN2, LDB3, NKX2-5, RBM20, TAFAZZIN, TRAF1, TRAF4)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Knochenmarkversagen (ACD, BRCA1, BRCA2, BRIP1, CTC1, DKC1, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, MECOM, NHP2, NOP10, PALB2, PARN, RAD51, RAD51C, RFWO3, RTEL1, SAMD9, SAMD9L, SLX4, SRP72, STN1, TERC, TERT, TINF2, TP53, UBE2T, UBE2V, UBE2W)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kolorektalkarzinom - Lynch-Syndrom (HNPCC) (EPCAM, MLH1, MSH2, MSH6, PMS2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kolorektalkarzinom - Polyposis (FAP) (APC, MUTYH)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kolorektalkarzinom - weitere Gene (AKT1, ATM, BMPR1A, CHEK2, GALNT12, GREM1, MLH3, MSH3, NTHL1, POLD1, POLE, PTEN, RNF43, RPS20, SCG5, SMAD4, STK11, TP53)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, assoziiert mit angeborener Thrombozytopenie (ARPC1B, WAS, WIPF1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, assoziiert mit anhidrotischen ektodermalen Dysplasien (IKKBK, IKBKG, NFKBIA)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, assoziiert mit immuno-ossären Dysplasien (EXTL3, MYSM1, RMRP, SMARCA11)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

Kombinierte Immundefekte - syndromal, assoziiert mit Thymusdefekten (CHD7, FOXN1, SEMA3E, TBX1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, Calciumkanaldefekte (CRACR2A, ORAI1, STIM1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, DNA-Reparaturdefekte (ATM, BLM, CDCA7, DNMT3B, GINS1, HELLS, LIG1, MCM10, MCM4, NBN, NSMCE3, PMS2, POLA1, POLE, POLE2, RNF168, ZBTB24)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, Hyper IgE Syndrom (HIES) (CARD11, DOCK8, ERBIN, IL6R, IL6ST, PGM3, SPINK5, STAT3, TGFBR1, TGFBR2, ZNF341)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, sonstige (BCL11B, CCBE1, CD28, DIAPH1, EPG5, ERCC6L2, FAT4, IKZF3, KDM6A, KMT2A, KMT2D, NFE2L2, PNP, RBCK1, RNF31, SKIC2, SKIC3, SP110, STAT5B, TTC7A)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierte Immundefekte - syndromal, Störungen des Vitamin-B12- und Folat-Stoffwechsels (MTHFD1, SLC46A1, TCN2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Kombinierter Immundefekt, weniger schwerwiegend (CID) (B2M, BCL10, CARD11, CD3G, CD40, CD40LG, CD8A, CHUK, CIITA, COPG1, DOCK2, DOCK8, FCHO1, ICOS, ICOSLG, IKKB, IKZF1, IKZF2, IL21, IL21R, ITK, LCK, MALT1, MAN2B2, MAP3K14, MSN, POLD1, POLD2, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, SASH3, STK4)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Komplementdefekte (C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C8G, C9, CD46, CD55, CD59, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, FCN3, MASP2, SERPING1, THBD)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Li-Fraumeni-Syndrom / Li-Fraumeni-ähnliche Symptomatik (TP53, CHEK2, POT1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Magenkarzinom (CDH1, CTNNA1, APC, ATM, BMPR1A, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, PTEN, SMAD4, STK11, TP53)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

männliche Infertilität - isoliert (ADGRG2, AR, AURKC, CFAP251, CFAP43, CFAP44, CFTR, DNAH1, DPY19L2, PLCZ1, SUN5, TEX11, ARMC2, CFAP65, CFAP69, CFAP91, DMRT1, DNAH17, FANCM, FSIP2, KLHL10, M1AP, MEI1, PMFBP1, QRICH2, SEPTIN12, SPEF2, Stag3, SYCP2, SYCP3, TEX14, TEX15, TSGA10, TTC29, USP26, USP91)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
männliche Infertilität - syndromal (APOA1, CATSPER2, CCDC39, CCDC40, CDC14A, CEP290, DNAAF11, DNAAF2, DNAAF4, DNAAF6, FANCA, MNS1, NLRP3, PKD1, RSPH3, SPEF2, TRIM37)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
männliche Infertilität – Syndrome des Reproduktionssystems / Endokrine Erkrankungen (AMH, AMHR2, ANOS1, AR, CHD7, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, FGFR1, GNRHR, HSD17B3, HSD3B2, KISS1R, LHB, LHCGR, NROB1, NR5A1, POU1F1, PROKR2, PROP1, SEMA3A, SOX10, SOX2, SRD5A2, SRY, TACR3, BMP4, BMP7, BNC2, CCDC141, DHX37, FGF17, FGF8, FSHB, FSHR, GATA4, GNRH1, HSGST1, IGSF10, IL17RD, INSL3, MAMLD1, MYRF, PLXNA1, PROK2, RSP01, SOX3, SOX9, STAR, WDR11,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Marfan-Syndrom / Marfan-ähnliche Erkrankungen (FBN1, ADAMTSL4, TGFB1, TGFB2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Melanom (BAP1, CDKN2A, MC1R, MITF, ACD, ATM, CDK4, POT1, PTEN, RB1, TERF2IP, TERT, TP53)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Morbus Osler (Hereditäre, hämorrhagische Teleangiectasie; HHT) (ACVRL1, ENG, SMAD4, BMPR2, EPHB4, GDF2, RASA1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Motilitätsstörungen (ACTB, CEBPE, CFTR, CTSC, FERMT3, FPR1, ITGB2, MRTFA, RAC2, SLC35C1, WDR1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Neuroendokrine Tumore (CDKN1B, MEN1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, VHL, AIP, APC, CDC73, DICER1, FH, MAX, NF1, PRKAR1A, PTEN, TMEM127, TP53, TSC1, TSC2, WRN)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Neurofibromatose / Schwannomatose (NF1, NF2, LZTR1, PRKAR1A, SMARCB1, SMARCE1, SPRED1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

<p>Nicht Inflammasom-bedingte Erkrankungen (ADAM17, ALPI, AP1S3, CARD14, COPA, HAVCR2, HCK, IKBKG, IL1RN, IL36RN, LPIN2, NCKAP1L, NOD2, OTULIN, PLCG2, PSMB8, PSMB9, PSMG2, PSTPIP1, SH3BP2, SLC29A3, SYK, TBK1, TNFAIP3, TNFRSF1A, TNFRSF22)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Nierenzellkarzinom (AKT1, ATM, BAP1, BRCA2, CDC73, CDKN2B, CHEK2, EPCAM, FH, FLCN, MAX, MET, MITF, MLH1, MSH2, MSH6, PIK3CA, PMS2, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SMARCA4, SMARCB1, TMEM127, TSC1, TSC2, VHL)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Noonan Syndrom / RASopathien (BRAF, KRAS, LZTR1, PTPN11, RAF1, RIT1, SOS1, CBL, HRAS, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, RRAS2, SHOC2, SOS2, SPRED1, SPRED2)</p>	<p>EDTA-Blut, Fruchtwasser, Chorionzotten, Zellkulturen (aus Fruchtwasser, Chorionzotten), DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Oligodontie, isoliert (AXIN2, EDA, EDAR, EDARADD, FGFR1, GREM2, IRF6, LRP6, LTBP3, MSX1, PAX9, PTH1R, SUMO1, TGFA, WNT10A, WNT10B)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Osteogenesis imperfecta / Frakturneigung / Osteoporose (COL1A1, COL1A2, AGA, ALPL, ANOS5, ANTXR2, ARHGAP25, ASXL1, ATP6V0A2, B3GALT6, B3GAT3, B4GALT7, BANF1, BMP1, CA2, CASR, CCDC134, CHST3, CLCN5, CLCN7, COL2A1, CORB2, CREB3L1, CRTAP, CTSK, CYP27B1, CYP2R1, EXOC6B, FGFR1, FKBP10, FN1, GBA1, GNAS, GNPTAB, GORAB, HRAS, IDH1, IDH2, IFIH1, IFITM5, IL6ST, KDELR2, LBR, LEMD3, LIFR, LPIN2, LRP5, LTBP1, MBTPS2, MESD, MET, MMP14, MMP2, MTAP, MYH3, NFIX, NOTCH2, NRAS, P3H1, P4HB, PLEKHM1, PLOD2, PLS3, POLR3A, PPIB, PRKACA, PRKAR1A, PTH1R, PYCR1, RECQL4, RIG1, RUNX2, SEC24D, SERPINF1, SERPINH1, SGMS2, SLC29A3, SLC34A3, SMS, SNX10, SOX9, SP7, SPARC, SQSTM1, TCIRG1, TENT5A, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TREM2, TRIP11, TRPV6, TYROBP, VCP, VDR, WNT1, WNT2)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080</p>
<p>Osteopetrose / Osteosklerose / erhöhte Knochendichte (ABCC9, ACP5, AMER1, ANKH, ANOS, BMP1, CA2, CLCN7, COL1A1, CSF1R, CTSK, DHCR24, DLX3, DMP1, DVL1, EBP, EIF2AK3, ENPP1, FAM111A, FAM20C, FERMT3, FGF23, FLNA, GALNT3, GBA1, GJA1, GNAS, HGSNAT, HHAT, HPGD, HSPG2, IKBKG, KL, LBR, LEMD3, LMNA, LPIN2, LRP4, LRP5, LRRK1, MAP2K1, MAP3K7, MTAP, NAGLU, OSTM1, PHEX, PLEKHM1, POLR3B, POLR3GL, PTDSS1, PTH1R, RASGRP2, RUNX2, SERP4, SGSH, SIK3, SLC26A2, SLC29A3, SLC4A2, SLCO2A1, SNX10, SOST, SP7, SQSTM1, TBCE, TBXAS1, TCIRG1, TGFB1, TMEM53, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TRAPPC2, TRPS1, TYROBP, VCP)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung</p>	<p>va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)</p>

Pankreaskarzinom (APC, ATM, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK1, STK11, TP53, VHL)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Pankreatitis (CLDN2, CPA1, CTRC, PRSS1, SPINK1, TRPV6, CFTR, CASR, CEL, CELA3B)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Peutz Jeghers Syndrom / Peutz-Jeghers-ähnliche Symptomatik (STK11, APC, BMPR1A, EPCAM, MLH1, MSH2, MSH6, PMS2, PTEN, SMAD4)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Phäochromozytom / Paragangliom-Pheochromozytom-Syndrom (MAX, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, DLST, EPAS1, FH, MDH2, MEN1, NF1, RET, SLC25A11, VHL)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Polyzystische Nierenerkrankungen (isoliert) (PKD1, PKD2, ALG5, ALG9, DNAJB11, DZIP1L, GANAB, HNF1B, IFT140, PKHD1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Prädisposition für invasive Pilzinfektionen (CARD9)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Prostatakarzinom (BRCA2, ATM, BRCA1, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, RAD51C, TP53)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Pulmonal-arterielle Hypertonie (PAH) (BMPR2, ABCC8, ACVRL1, ATP13A3, CAV1, EIF2AK4, ENG, GDF2, GGX, KCNK3, KDR, SMAD9, SOX17, TBX4, TET2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Regulatorische T-Zell-Defekte (BACH2, CTLA4, DEF6, FERMT1, FOXP3, IKZF1, IL2RA, IL2RB, LRBA, STAT3)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Retinoblastom (RB1, IKBKG, NDP, TSC1, TSC2, VHL)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080
Schilddrüsenkarzinom (AKT1, APC, CHEK2, DICER1, FOXE1, PRKAR1A, PTEN, RET, TP53, WRN)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-002-00b-8-2-015-080

Schwerer kombinierter Immundefekt (SCID), T-B- SCID (ADA, AK2, DCLRE1C, LIG4, NHEJ1, PRKDC, RAC2, RAG1, RAG2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Schwerer kombinierter Immundefekt (SCID), T-B+ SCID (CD247, CD3D, CD3E, CORO1A, IL2RG, IL7R, ITPKB, JAK3, LAT, LCP2, PAX1, PTPRC)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Schwerhörigkeit (GJB2, GJB6, ABCC1, ABHD12, ACOX1, ACTB, ACTG1, ADCY1, ADGRV1, AFG2A, AFG2A, AFG2B, AFG2B, AIFM1, ALMS1, AMMECR1, ANKH, AP1S1, ATOH1, ATP11A, ATP2B2, ATP6V0A4, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTBD, CABP2, CACNA1D, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CEMIP, CEP250, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, CLRN2, COCH, COG4, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRLS1, CRYM, DCAF17, DCDC2, DIABLO, DIAPH1, DIAPH3, DLX1, DMXL2, DNAJC3, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPHA10, EPS8, EPS8L2, ERAL1, ESPN, ESRP1, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR1, FGFR2, FGFR3, FITM2, FOXF2, FOXI1, FOXL1, GAB1, GAS2, GATA3, GDF6, GGPS1, GIPC3, GJA1, GJB3, GOSR2, GPR156, GPRASP2, GPSM2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, GSDME, HAAO, HARS2, HGF, HOMER2, HOXA2, HOXB1, HSD17B4, IFNL1, ILDR1, KARS1, KCNE1, KCNJ10, KCNJ16, KCNQ1, KCNQ4, KDM3B, KIT, KITLG, KMT2D, LARS2, LETM1, LHFPL5, LHX3,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
solide Tumore bei Erwachsenen (Gesamtpanel) (ABRAXAS1, ACD, AIP, AKT1, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CHEK2, CTNNA1, CTR9, DICER1, DIS3L2, DLST, ELP1, EPAS1, EPCAM, ERCC4, FAAP100, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBXW7, FH, FLCN, FOXE1, GALNT12, GPC3, GPR161, GREM1, HOXB13, IKBKG, KIT, LZTR1, MAD2L2, MAX, MC1R, MDH2, MEN1, MET, MITF, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NDP, NF1, NF2, NTHL1, NYNRIN, PALB2, PDGFRA, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POT1, POU6F2, PRKAR1A, PRSS1, PTCH1, PTEN, RAD51, RAD51C, RAD51D, RB1, RECQL4, REST, RET, RFWF3, RNF43, RPS20, SCG5, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, SPRED1, STK11, SUFU, TERF2IP, TERT, TMEM127, TOP3A, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRN, WT1, XRCC2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

Sonstige nichtlymphoide Defekte (CSF2RA, CSF2RB, GATA2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Sphärozytose (ANK1, SLC4A1, SPTB, EPB42, SPTA1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Störungen der Knochenmineralisierung (ALPL, ANKH, CASR, CDC73, CLCN5, CYP27B1, CYP2R1, CYP3A4, DMP1, ENPP1, FGF23, GCM2, HNRNPC, HRAS, NRAS, PHEX, SGK3, SLC34A3, TNFRSF11B, TRPV6, VDR)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Thrombozytopenie (ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CDC42, CYCS, DIAPH1, ETV6, FLI1, FLNA, FYB1, GALE, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP9, HOXA11, IKZF5, ITGA2B, ITGB3, KDSR, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, PLAU, PRKACG, PTPN11, PTPRJ, RBM8A, RUNX1, SLC35A1, SLFN14, SRC, STIM1, THPO, TPM4,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
TLR-Signalwegdefizit mit bakterieller Anfälligkeit (IRAK1, IRAK4, MYD88, TIRAP, TLR8)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Tumor des zentralen Nervensystems / Medulloblastom (AIP, ALK, APC, ATM, BRCA2, CDKN2A, CHEK2, DICER1, ELP1, GPR161, LZTR1, MLH1, MSH2, MSH6, NF1, NF2, PHOX2B, PMS2, POT1, PTCH1, PTEN, SMARCA4, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Typ-1-Interferonopathien (ACP5, ADA2, ADAR, ATAD3A, C2orf69, CDC42, DNASE1L3, DNASE2, IFIH1, LSM11, OAS1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, STAT2, STING1, TREX1, USP18)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Variablen Immundefektsyndrom (CVID) / Hypogammaglobulinämie (ARHGEF1, ATP6AP1, CD19, CD81, CR2, IKZF1, IRF2BP2, MOGS, MS4A1, NFKB1, NFKB2, PIK3CD, PIK3CG, PIK3R1, POU2AF1, PTEN, RAC2, SEC61A1, SH3KBP1, TNFRSF13B, TNFRSF13C, TNFRSF13D, TNFRSF14)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Vaskuläre Fehlbildungen (inkl. HTT, AVM, CM-AVM, FCCM) (ACVRL1, CCM2, ENG, KRIT1, PDCD10, PTEN, RASA1, RASA1, SMAD4, BMPR2, ELMO2, EPHB4, GDF2, GLMN, NOTCH3, PIK3CA, SOX18, STAMBP, TEK)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080

Very-Early-Onset-Inflammatory-Bowel-Disease (VEO-IBD) (ADA, ADAM17, AICDA, ALPI, ANKZF1, ANO1, BACH2, BTK, CARMIL2, CD3G, CD40LG, CTLA4, CYBA, CYBB, CYBC1, DCLRE1C, DKC1, DOCK11, DOCK8, DUOX2, EPCAM, FERMT1, FOXP3, G6PC3, GUCY2C, HPS1, HPS4, HPS6, ICOS, IFIH1, IKBKKG, IL10, IL10RA, IL10RB, IL21, IL2RA, IL2RB, IL2RG, ITGB2, LIG4, LRBA, MEFV, MVK, NCF1, NCF2, NCF4, NLRCA, NOD2, NOX1, NPC1, OTULIN, PI4KA, PIK3CD, PIK3R1, PLCG2, RAG1, RAG2, RELA, RIPK1, RTEL1, SAMD9, SH2D1A, SKIC2, SKIC3, SLC37A4, SLC9A3, SLC02A1, SOCS1, STAT1, STAT3, STXBP2, STXBP3, TGFBR1, TGFBR1, TGFBR2, TNFAIP3, TRIM22, TTC7A, WAS, XIAP, ZAP70)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Wilms-Tumor (WT1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CTR9, DICER1, DIS3L2, FBXW7, GPC3, NYNRRN, PALB2, POU6F2, REST, TP53, TRIM28, TRIM37, TRIP13)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
Ziliendyskinesie, primäre (CCDC103, CCDC39, CCDC40, DNAH11, DNAH5, DNAI1, ODAD2, ODAD3, SPAG1, ZMYND10, CCDC65, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH9, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, ODAD1, ODAD4, OFD1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_IDT (07.03.2025) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022) varfeed-varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.4.1-fc9-093-00b-8a2-015-080
BCR::ABL, Mutationsanalyse (CML, ALL)	EDTA-Blut, EDTA-Knochenmark; RNA	PCR, Sanger-Sequenzierung	va_Mutationsanalyse_BCR-ABL1 (14.10.2022)
BCR::ABL, qualitativer Nachweis (CML, ALL)	EDTA-Blut, EDTA-Knochenmark; RNA	PCR, Gelelektrophorese	va_Qualitative_BCR-ABL1_PCR_Multiplex (14.10.2022)
BCR::ABL, quantitativ (CML, ALL)	EDTA-Blut, EDTA-Knochenmark; RNA	PCR, Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR)	va_Quantitative_BCR-ABL1_PCR_QS5 (12.10.2022)

## Untersuchungsgebiet: Transfusionsmedizin

### Untersuchungsart:

#### Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
HLA-Klasse I, II	Blut (EDTA, Citrat), DNA; DNA	PCR (SSP) / Gelelektrophorese	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektrophorese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022)
HLA-Klasse I, II	Blut (EDTA, Citrat), DNA; DNA	qPCR (SSP)	tpi_aa_Rainbow (24.10.2022) tpi_aa_B27 (24.10.2022)
HLA-Klasse I, II	Blut (EDTA, Citrat), DNA; DNA	PCR, NGS; sequence capture, sequencing-by-synthesis Next-Generation-Sequencing	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
Thrombozytenantigene (HPA), molekulargenetisch	Blut (EDTA, Citrat), DNA; DNA	qPCR (SSP)	tpi_aa_qPCR_ERYQ.doc (24.10.2022)