## Untersuchungsgebiet: Humangenetik

**Untersuchungsart:**
**Chromosomenanalyse**

| Analyt (Messgröße) | Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial) | Untersuchungstechnik | Anweisung+Version Pipeline/Kit/Panel+Version |
|---|---|---|---|
| Angeborener Chromsomensatz | Blut, Fruchtwasser, Chorionzotten oder andere Gewebeproben[a] | Chromosomenbänderungsanalyse | va_Bearb_peripheres_Blut (15.09.2026)<br>va_Bearb_Fruchtwasser (15.09.2026)<br>va_Bearb_Chorionzotten (15.09.2026)<br>va_Bearb_Fibroblasten (15.09.2026) |
| Angeborener Chromsomensatz | Blut, Fruchtwasser, Chorionzotten oder andere Gewebeproben[a], Mundschleimhautabstrich | Fluoreszenz-in-situ-Hybridisierung (FISH) | va_Bearb_peripheres_Blut (15.09.2026)<br>va_Bearb_Fruchtwasser (15.09.2026)<br>va_Bearb_Chorionzotten (15.09.2026)<br>va_Bearb_Fibroblasten (15.09.2026)<br>aa_FISH_Mundschleimhautabstrich (15.09.2026) |
| Erworbener Chromosomensatz | Blut, Knochenmark | Chromosomenbänderungsanalyse | va_Bearb_peripheres_Blut (15.09.2026)<br>aa_Probenannahme_u_Ansatz_TG (15.09.2026)<br>Praeparatherst_TG (15.09.2026)<br>aa_Karyotypanalyse_TG (15.09.2026) |
| Erworbener Chromosomensatz | Blut, Knochenmark, Knochenmarkausstrich | Fluoreszenz-in-situ-Hybridisierung (FISH) | va_Bearb_peripheres_Blut (15.09.2026)<br>va_Bearb_Fruchtwasser (15.09.2026)<br>va_Bearb_Chorionzotten (15.09.2026)<br>va_Bearb_Fibroblasten (15.09.2026)<br>aa_FISH_Mundschleimhautabstrich (15.09.2026)<br>aa_FISH_KM_Ausstrich (15.09.2026)<br>aa_FISH_KM_u_Blut_aus_Kultur (15.09.2026) |
| Angeborener Chromosomensatz | Blut, DNA; DNA | molekulare Karyotypisierung mittels Array-CGH | va_Array_CGH (12.05.2025) |

[a]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 |
|---|---|---|---|
| Angelmann-Syndrom (15q11-q13 Region) | EDTA-Blut, DNA; DNA | MLPA (methylierungssensitiv) | va_MS_MLPA_PWAS (16.09.2026) |
| Prader-Willi-Syndrom (15q11-q13 Region) | EDTA-Blut, DNA; DNA | MLPA (methylierungssensitiv) | va_MS_MLPA_PWAS (16.09.2026) |
| Ausschluss/ Nachweis mütterlicher Zellen im pränatalen Untersuchungsmaterial bzw. Abortdiagnostik | EDTA-Blut, Fruchtwasser, Chorionzotten oder andere Gewebeproben[a], Zellkulturen (aus Fruchtwasser, Chorionzotten und anderen Gewebeproben[a]), DNA; DNA | PCR, Fragmentanalyse | va_Ablauf_muetterliche_Kontamination (16.09.2026) |
| pränataler Schnelltest (STR-basiert) | EDTA-Blut, Fruchtwasser, Chorionzotten oder andere Gewebeproben[a], Zellkulturen (aus Fruchtwasser, Chorionzotten und anderen Gewebeproben[a]), DNA; DNA | PCR, Fragmentanalyse | va_STR_Praenataler_Schnelltest (16.09.2026) |
| 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 (16.09.2026) |
| 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 (09.09.2026) |
| Azoospermie (Deletionen im AZF-Locus: AZFa, AZFb, AZFc) | EDTA-Blut, DNA; DNA | SSP-PCR, Fragmentanalyse | va_AZF_Diagnostik (16.09.2026) |

| | | | |
|---|---|---|---|
| Fragiles-X-Syndrom (FMR1) | EDTA-Blut, DNA; DNA | PCR, Triplet Repeat-primed PCR, Fragmentanalyse | va_FRAX.docx (09.05.2025) |
| Chorea Huntington (Bestimmung der CAG/CCG-Repeats im HTT-Gen) | EDTA-Blut, DNA; DNA | Fragmentanalyse | va_HD (20.10.2025) |
| DPYD-Defizienz / 5-FU-Toxizität (DPYD: dbSNP rs3918290, rs55886062, rs67376798, rs56038477) | EDTA-Blut, DNA; DNA | Loop Amplification, Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR) | va_LAMP_DPYD (02.07.2025) |
| Whole Exome (WES) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Whole Genome (WGS) | EDTA-Blut, DNA; DNA | NGS (PCR-free library, sequencing-by-synthesis, custom pipeline, SNV, indel, CNV) MLPA, Sanger-Sequenzierung | ukj_ih_va_WGS 26.5.2025<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Amelogenesis imperfecta (syndromal) (CNNM4, FAM20C, LTBP3, ORAI1, PEX1, PEX26, PEX6, ROGDI, SLC10A7, SLC13A5, STIM1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Andere angeborene Immunitätsstörungen im Zusammenhang mit nicht hämatopoetischen Geweben (APOL1, CLCN7, HMOX1, NBAS, NCSTN, OSTM1, PLEKHM1, PSEN1, PSENEN, RANBP2, RPSA, SNX10, TCIRG1, TNFRSF11A, TNFSF11) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Anfälligkeit für schwere Virusinfektion (FCGR3A, IFIH1, IFNAR1, IFNAR2, IRF7, IRF9, NOS2, POLR3A, POLR3C, POLR3F, STAT1, STAT2, TLR7, ZNFX1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Angeborene Neutropenie (CEBPE, CLPB, CSF3R, CXCR2, DNAJC21, EFL1, ELANE, G6PC3, GFI1, HAX1, HYOU1, JAGN1, LAMTOR2, SBDS, SLC37A4, SMARCD2, SRP54, TAFAZZIN, USB1, VPS13B, VPS45, WAS) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Angioödem (SERPING1, ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Autoimmunität mit oder ohne Lymphoproliferation (AIRE, ITCH, JAK1, PDCD1, PEPD, SOCS1, TPP2) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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, ZNF469) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Brust- und Eierstockkrebs (ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, NBN, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Cowden-Syndrom / Cowden-Syndrom-ähnliche Symptomatik (PTEN, AKT1, PIK3CA, SDHB, SDHC, SDHD, SEC23B) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |

| | | | |
|---|---|---|---|
| Cystische Fibrose (CFTR) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Defekte des Inflammasoms (MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, RIPK1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Familiäres thorakales Aortenaneurysma und Aortendissektion (TAAD) (ACTA2, FBN1, FOXE3, LOX, SMAD3, TGFBR1, TGFBR2, 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, TGFB2, TGFB3, THSD4) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, RFWD3, SLX4, TOP3A, UBE2T, XRCC2) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Hereditäre Neuropathien (HMSN/HNPP/CMT) (GDAP1, GJB1, HINT1, MFN2, MPZ, PMP22, SH3TC2, SORD, AARS1, ABCA1, AIFM1, ARHGEF10, ATL1, ATL3, 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, WARS1, WNK1, YARS1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Hereditäre Schmerzerkrankungen (CIP, SFN, PE, PEPD, FEPS1-3, FD) / HSAN (KIF1A, RETREG1, SCN9A, WNK1, ATL1, ATL3, DNMT1, DST, ELP1, FLVCR1, GLA, NGF, NTRK1, PRDM12, RAB7A, SCN10A, SCN11A, SPTLC1, SPTLC2, TECPR2, TRPA1, TTR, ZFHX2) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Hereditäre Spastische Paraplegie (HSP, SPG) (ATL1, 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, BTD, 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) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Hermansky-Pudlak-Syndrom (DD Chediak-Higashi-Syndrom, Griscelli-Syndrom) (AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, RAB27A) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |

| | | | |
|---|---|---|---|
| Herpes-simplex-Enzephalitis (HSE) (ATG4A, DBR1, IRF3, MAP1LC3B2, SNORA31, TBK1, TICAM1, TLR3, TRAF3, UNC93B1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Herzrhythmusstörung - Erregungsleitungsstörungen / Sick-Sinus-Syndrom / Brugada-Syndrom (CCD, PCCD, SSS, SND, BRU) (CACNA1D, CASQ2, DES, DMD, EMD, GJC1, GLA, GNB2, GNB5, 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Hyper-IgM-Syndrom (AICDA, CTNNBL1, 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, TBX5, TPM1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, RFWD3, RTEL1, SAMD9, SAMD9L, SLX4, SRP72, STN1, TERC, TERT, TINF2, TP53, UBE2T, WRAP53, XRCC2) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Kombinierte Immundefekte - syndromal, assoziiert mit anhidrotischen ektodermalen Dysplasien (IKBKB, IKBKG, NFKBIA) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Kombinierte Immundefekte - syndromal, assoziiert mit immuno-ossären Dysplasien (EXTL3, MYSM1, RMRP, SMARCAL1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, IKBKB, IKZF1, IKZF2, IL21, IL21R, ITK, LCK, MALT1, MAN2B2, MAP3K14, MSN, POLD1, POLD2, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, SASH3, STK4, TAP1, TAP2, TAPBP, TFRC, TNFRSF4, ZAP70) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, XRCC2) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Marfan-Syndrom / Marfan-ähnliche Erkrankungen (FBN1, ADAMTSL4, TGFBR1, TGFBR2) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Morbus Osler (Hereditäre, hämorrhagische Teleangiektasie; 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |

| | | | |
|---|---|---|---|
| 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, TRIM22) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Noonan Syndrom / RASopathien (BRAF, KRAS, LZTR1, PTPN11, RAF1, RIT1, SOS1, CBL, HRAS, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, RRAS2, SHOC2, SOS2, SPRED1, SPRED2) | EDTA-Blut, Fruchtwasser, Chorionzotten, Zellkulturen (aus Fruchtwasser, Chorionzotten), DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Oligodontie, isoliert (AXIN2, EDA, EDAR, EDARADD, FGFR1, GREM2, IRF6, LRP6, LTBP3, MSX1, PAX9, PTH1R, SUMO1, TGFA, WNT10A, WNT10B) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Osteogenesis imperfecta / Frakturneigung / Osteoporose (COL1A1, COL1A2, AGA, ALPL, ANO5, ANTXR2, ARHGAP25, ASXL1, ATP6V0A2, B3GALT6, B3GAT3, B4GALT7, BANF1, BMP1, CA2, CASR, CCDC134, CHST3, CLCN5, CLCN7, COL2A1, COPB2, 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, RIGI, 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, XYLT2) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Osteopetrose / Osteosklerose / erhöhte Knochendichte (ABCC9, ACP5, AMER1, ANKH, ANO5, 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, SFRP4, SGSH, SIK3, SLC26A2, SLC29A3, SLC4A2, SLCO2A1, SNX10, SOST, SP7, SQSTM1, TBCE, TBXAS1, TCIRG1, TGFB1, TMEM53, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TRAPPC2, TRPS1, TYROBP, VCP) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026) |

| | | | |
|---|---|---|---|
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Prädisposition für invasive Pilzinfektionen (CARD9) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Pulmonal-arterielle Hypertonie (PAH) (BMPR2, ABCC8, ACVRL1, ATP13A3, CAV1, EIF2AK4, ENG, GDF2, GGCX, 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |

| | | | |
|---|---|---|---|
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, BTD, 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, IFNLR1, 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, RFWD3, 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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, TRPM7, TUBB1, WAS, WIPF1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Variables Immundefektsyndrom (CVID) / Hypogammaglobulinämie (ARHGEF1, ATP6AP1, CD19, CD81, CR2, IKZF1, IRF2BP2, MOGS, MS4A1, NFKB1, NFKB2, PIK3CD, PIK3CG, PIK3R1, POU2AF1, PTEN, RAC2, SEC61A1, SH3KBP1, TNFRSF13B, TNFRSF13C, TNFSF12, TRNT1) | EDTA-Blut, DNA; DNA | NGS (sequence capture, sequencing-by-synthesis, custom pipeline, SNV, indel) MLPA, Sanger-Sequenzierung | va_Sanger (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| Wilms-Tumor (WT1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CTR9, DICER1, DIS3L2, FBXW7, GPC3, NYNRIN, 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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-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 (16.09.2026)<br>va_NGS_IDT (12.05.2025)<br>va_MLPA (12.05.2025)<br>va_digitale_MLPA (17.09.2026)<br>varfeed- varvis (Limbus Medical Technologies GmbH) varfeed- Version 2.11.0-fc9-093-00b-8a2-015-080 |
| BCR::ABL, Mutationsanalyse (CML, ALL) | EDTA-Blut, EDTA-Knochenmark; RNA | PCR, Sanger-Sequenzierung | va_Mutationsanalyse_BCR-ABL1 (22.06.2026) |
| BCR::ABL, qualitativer Nachweis (CML, ALL) | EDTA-Blut, EDTA-Knochenmark; RNA | PCR, Gelelektrophorese | va_Qualitative_BCR-ABL1_PCR_Multiplex (22.06.2026) |
| BCR::ABL, quantitativ (CML, ALL) | EDTA-Blut, EDTA-Knochenmark; RNA | PCR, Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR) | va_Quantitative_BCR-ABL1_PCR_QS5 (01.07.2026) |

## 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 (16.09.2026)<br>tpi_aa_PRA_Agarosegele (16.09.2026)<br>tpi_aa_Elektropherese (16.09.2026)<br>tpi_aa_HLA_SSP_HSP (17.09.2026) |
| HLA-Klasse I, II | Blut (EDTA, Citrat), DNA; DNA | qPCR (SSP) | tpi_aa_Rainbow (16.09.2026)<br>tpi_aa_B27 (16.09.2026) |

| | | | |
|---|---|---|---|
| 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 (16.09.2026) |
| Thrombozytenantigene (HPA), molekulargenetisch | Blut (EDTA, Citrat), DNA; DNA | qPCR (SSP) | tpi_aa_qPCR_ERYQ.doc (16.09.2026) |