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) va_Bearb_Fruchtwasser (15.09.2026) va_Bearb_Chorionzotten (15.09.2026) 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) va_Bearb_Fruchtwasser (15.09.2026) va_Bearb_Chorionzotten (15.09.2026) va_Bearb_Fibroblasten (15.09.2026) aa_FISH_Mundschleimhautabstrich (15.09.2026)
Erworbener Chromosomensatz	Blut, Knochenmark	Chromosomenbänderungsanalyse	va_Bearb_peripheres_Blut (15.09.2026) aa_Probenannahme_u_Ansatz_TG (15.09.2026) Praeparatherst_TG (15.09.2026) aa_Karyotypanalyse_TG (15.09.2026)
Erworbener Chromosomensatz	Blut, Knochenmark, Knochenmarkausstrich	Fluoreszenz-in-situ-Hybridisierung (FISH)	va_Bearb_peripheres_Blut (15.09.2026) va_Bearb_Fruchtwasser (15.09.2026) va_Bearb_Chorionzotten (15.09.2026) va_Bearb_Fibroblasten (15.09.2026) aa_FISH_Mundschleimhautabstrich (15.09.2026) aa_FISH_KM_Ausstrich (15.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) 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 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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) va_NGS_IDT (12.05.2025) va_MLPA (12.05.2025) va_digitale_MLPA (17.09.2026) 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)