Untersuchungsgebiet: Humangenetik

Untersuchungsart:
Chromosomenanalyse**

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version
Angeborener Chromsomensatz	Blut, Fruchtwasser, Gewebeproben (Chorion, Abortmaterial, Fibroblasten)[a]	Chromosomenbänderungsanalyse	va_Bearb_peripheres_Blut (07.04.2022) va_Bearb_Fruchtwasser (11.02.2022) va_Bearb_Chorionzotten (01.12.2023) va_Bearb_Fibroblasten (07.04.2022)
Angeborener Chromsomensatz	Blut, Fruchtwasser, Gewebeproben (Chorion, Abortmaterial, Fibroblasten, Mundschleimhaut)[a]	Fluoreszenz-in-situ-Hybridisierung (FISH)	va_Bearb_peripheres_Blut (07.04.2022) va_Bearb_Fruchtwasser (11.02.2022) va_Bearb_Chorionzotten (01.12.2023) va_Bearb_Fibroblasten (07.04.2022) aa_FISH_Mundschleimhautabstrich (01.12.2023)
Erworbener Chromosomensatz	Blut, Knochenmark	Chromosomenbänderungsanalyse	va_Bearb_peripheres_Blut (07.04.2022) aa_Probenannahme_u_Ansatz_TG (01.12.2023) Praeparatherst_TG (23.10.2023) aa_Karyotypanalyse_TG (01.12.2023)
Erworbener Chromosomensatz	Blut, Knochenmark, Knochenmarkausstrich	Fluoreszenz-in-situ-Hybridisierung (FISH)	va_Bearb_peripheres_Blut (07.04.2022) va_Bearb_Fruchtwasser (11.02.2022) va_Bearb_Chorionzotten (01.12.2023) va_Bearb_Fibroblasten (07.04.2022) aa_FISH_Mundschleimhautabstrich (01.12.2023) aa_FISH_KM_Ausstrich (13.04.2022) aa_FISH_KM_u_Blut_aus_Kultur (13.04.2023)
Angeborener Chromosomensatz	Blut, DNA; DNA	molekulare Karyotypisierung mittels Array-CGH	va_Array_CGH (26.10.2022)

[a]Die Proben werden vom Labor ohne vorherige histologische Beurteilung bearbeitet und analysiert.

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

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 (23.10.2022)
Prader-Willi-Syndrom (15q11-q13 Region)	EDTA-Blut, DNA; DNA	MLPA (methylierungssensitiv)	va_MS_MLPA_PWAS (23.10.2022)
Ausschluss/ Nachweis mütterlicher Zellen im pränatalen Untersuchungsmaterial bzw. Abortdiagnostik	EDTA-Blut, Fruchtwasser, Chorionzotten, Gewebeprobena, Zellkulturen (aus Fruchtwasser, Chorionzotten und anderen Gewebeprobena), DNA; DNA	PCR, Mikrosatellitenanalyse	va_Ablauf_muetterliche_Kontamination (22.10.2022)
pränataler Schnelltest (STR-basiert)	EDTA-Blut, Fruchtwasser, Chorionzotten, Gewebeprobena, Zellkulturen (aus Fruchtwasser, Chorionzotten und anderen Gewebeprobena), DNA; DNA	PCR, Mikrosatellitenanalyse	va_STR_Praenataler_Schnelltest (28.09.2022)
Uniparentale Disomien der Chromosomen 6, 7, 11, 13, 14, 15, 18, 21	EDTA-Blut, Fruchtwasser, Chorionzotten, Zellkulturen (aus Fruchtwasser, Chorionzotten), DNA; DNA	PCR, Mikrosatellitenanalyse	va_MK_UPD_Abort_CE (21.10.2022)
Cystische Fibrose: Screening der häufigsten Mutationen (CFTR)	EDTA-Blut, Fruchtwasser, Chorionzotten, Zellkulturen (aus Fruchtwasser, Chorionzotten), DNA; DNA	SSP-PCR, Fragmentanalyse (CFTR-Assay Fa. Elucigene)	va_CFTR_DE (22.10.2022)
Azoospermie (Deletionen im AZF-Locus: AZFa, AZFb, AZFc)	EDTA-Blut, DNA; DNA	SSP-PCR, Fragmentanalyse	va_AZF_Diagnostik (21.10.2022)
Chorea Huntington (Bestimmung der CAG/CCG-Repeats im HTT-Gen)	EDTA-Blut, DNA; DNA	PCR, Short Tandem Repeat typing	va_Chorea_Huntington (21.10.2022)
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 (15.10.2021)

Retinoblastom (RB1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022)
Adenomatöse Polyposis coli (APC , MUTYH)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Herzrhythmusstörung - Katecholaminerge Polymorphe Ventrikuläre Tachykardie (CPVT) (CASQ2, RYR2, CALM1, CALM2, CALM3, KCNJ2, TECRL, TRDN)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Herzrhythmusstörung - Short-QT-Syndrom (SQTS) (KCNH2, KCNJ2, KCNQ1, SLC4A3)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)

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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MYO6, NRAS, PRKAG2, PTPN11, RAF1, RASA2, RIT1, RRAS, SLC25A4, SOS1, SOS2, TNNC1,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Marfan-Syndrom/ Marfan-ähnliche Erkrankungen (FBN1, ADAMTSL4, TGFBR1, TGFBR2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Morbus Osler (Hereditäre, hämorrhagische Teleangiektasie; HHT) (ACVRL1, ENG, SMAD4, BMPR2, EPHB4, GDF2, RASA1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)

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,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
männliche Infertilität - Syndrome des Reproduktionssystems / Endokrine Erkrankung (AMH, AMHR2, ANOS1, AR, CHD7, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, FGFR1, GNRHR, HSD17B3, HSD3B2, KISS1R, LHB, LHCGR, NR0B1, NR5A1, POU1F1, PROKR2, PROP1, SEMA3A, SOX10, SOX2, SRD5A2, SRY, TACR3, BMP4, BMP7, BNC2, CCDC141, DHX37, FGF17, FGF8, FSHB, FSHR, GATA4, GNRH1, HS6ST1, IGSF10, IL17RD, INSL3, MAMLD1, MYRF, PLXNA1, PROK2, RSPO1, SOX3, SOX9, STAR, WDR11,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)

Hereditäre Neuropathie (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, [illegible])	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Hereditäre Spastische Paraplegie (HSP, SPG) (ATL1, CYP7B1, HSPD1, KIF1A, REEP1, SPAST, SPG11, 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, [illegible])	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Polyzystische Nierenerkrankungen (isoliert) (PKD1, PKD2, ALG5, ALG9, DNAJB11, DZIP1L, GANAB, HNF1B, IFT140, PKHD1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)

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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Amelogenesis imperfecta (syndromal) (CNNM4, FAM20C, LTBP3, ORAI1, PEX1, PEX26, PEX6, ROGDI, SLC10A7, SLC13A5, STIM1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)

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, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Cowden-Syndrom (PTEN)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Cystische Fibrose: Komplettanalyse (CFTR)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_CFTR_DE (22.10.2022) va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022)
Li-Fraumeni-Syndrom/ Li-Fraumeni-like Syndrom (TP53, CHEK2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Lynch-Syndrom/ HNPCC (EPCAM, MLH1, MSH2, MSH6, PMS2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Melanom, familär (ASIP, BAP1, BRCA1, BRCA2, CCND1, CDK4, CDKN2A, EPCAM, MITF, MLH1, MSH2, MSH6, NF1, OCA2, PLA2G6, PMS2, PTEN, RAC1, RB1, SLC45A2, TERT)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Neurofibromatose (NF1, NF2)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022)
Pankreaskarzinom (APC, ATM, BRCA1, BRCA2, CDKN2A, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Peutz-Jeghers-Syndrom (STK11)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Darmkrebs/Weitere Gene (BMPR1A, EXO1, GALNT12, MLH3, NTHL1, PMS1, POLD1, POLE, PTEN, SMAD4, STK11, TP53)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Fanconi Anämie (BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCG, BRCA1, FANCF, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
Pankreatitis (CASR, CFTR, CTRC, PRSS1, SPINK1)	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)

Immundefekte (ACP5, ACTB, ADA, ADAR, AICDA, AIRE, AK2, AOAH, AP3B1, APOL1, ATM, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C3, C4A, C4B, C4BPA, C5, C6, C7, C8A, C8B, C9, CARD14, CARD9, CASP10, CASP8, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDH7, CEBPE, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, CHD7, CIITA, CLEC7A, COLEC11, CR2, CSF2RA, CSF3R, CTLA4, CTSC, CXCR4, CYBA, CYBB, DCLRE1C, DGCR2, DGCR8, DKC1, DNM2, DNMT3B, DOCK8, DTNBP1, ELANE, ESS2, F12, FADD, FANCA, FANCE, FAS, FASLG, FCGR3A, FCGR3B, FCN3, FERMT3, FGF10, FGFR2, FOXN1, FOXP3, FPR1, G6PC1, G6PC3, G6PD, GATA2, GFI1, HAX1, HFE, HPS1, HPS3, HPS4, HPS5, HPS6, ICOS, IFIH1, IFNGR1, IFNGR2, IGLL1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL1RN, IL21, IL21R, IL2RA, IL2RG, IL36RN, IL7R, IRAK4, IRF8, ITCH, ITGA3, ITGB2, ITK, JAK2, JAK3, KRAS, LAMTOR2, LIG1, LIG4, LPIN2, LRRC8A, LYST, MAGT1, MASP1, MASP2, MBL2, MCM4, MEFV, MLH1, MLPH, MPO, MRE11, MS4A1,	EDTA-Blut, DNA; DNA	NGS (sequence capture, sequencing-by-synthesis, varvis pipeline (Limbus Medical Technologies GmbH), SNV, indel) MLPA, Sanger-Sequenzierung	va_Sanger (20.10.2022) va_NGS_Keimbahn (20.10.2022) va_MLPA (23.10.2022) va_digitale_MLPA (21.06.2022)
BCR::ABL, Mutationsanalyse (CML, ALL)	EDTA-Blut, EDTA-Knochenmark; RNA	PCR, Sanger-Sequenzierung	va_Mutationsanalyse_BCR-ABL1 (14.10.2022)
BCR::ABL, qualitativer Nachweis (CML, ALL)	EDTA-Blut, EDTA-Knochenmark; RNA	PCR, Gelelektrophorese	va_Qualitative_BCR-ABL1_PCR_Multiplex (14.10.2022)
BCR::ABL, quantitativ (CML, ALL)	EDTA-Blut, EDTA-Knochenmark; RNA	PCR, Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR)	va_Quantitative_BCR-ABL1_PCR_QS5 (12.10.2022)

Untersuchungsgebiet: Immunologie

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
HLA-A-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
HLA-B-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
HLA-C-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
HLA-DRB1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
HLA-DRB3/4/5-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
HLA-DQB1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)

HLA-DQA1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
HLA-DPB1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
HLA-DPA1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_Rainbow (24.10.2022)
Thrombozytenantigene (HPA), molekulargenetisch	Blut (EDTA, Citrat) / genomische DNA	qPCR (SSP)	tpi_aa_qPCR_ERYQ.doc (24.10.2022)
Einzelantigene molekulargenetisch B*27; B*57:01	Blut (EDTA, Citrat) / genomische DNA	PCR (SSP)/ Gelelektrophorese / qPCR (SSP)	tpi_aa_HLA_SSP_Olerup (24.10.2022) tpi_aa_PRA_Agarosegele (24.10.2022) tpi_aa_Elektropherese (24.10.2022) tpi_aa_HLA_SSP_HSP (24.10.2022) tpi_aa_B27 (24.10.2022)
HLA-DRB1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-DQB1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-DRB3-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-DRB4-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-DRB5-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-A-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-B-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-C-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-DPB1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-DQA1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
HLA-DPA1-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)
MICA/MICB-Locus	Blut (EDTA, Citrat) / genomische DNA	PCR, NGS: sequence capture, sequencing-by-synthesis	tpi_aa_NGS_basierte_HLA_Typisierung (24.10.2022)