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Ordensklinikum Linz

Labor für
Molekulargenetische
Diagnostik

Genlisten Paneldiagnostik (Zuweiser Information)

Die folgenden Parameterlisten dienen zur Orientierung für Zuweiser und bilden nicht den gesamten Akkreditierungsumfang der Verfahren des Labors ab!

Multiplex PCR (Endpunkt-PCR Humangenomanalyse aus isolierten Nukleinsäuren) letzte Überarbeitung 10.03.2025

KMT2A::FOXO4, t(X;11)(q13;q23)
STIL, SIL::TAL1
KMT2A::MLLT11, t(1;11)(q21;q23)
KMT2A::EPS15, t(1;11)(p32;q23)
TCF3::PBX1, t(1;19)(q23;p13)
NPM1::MLF1, t(3;5)(q25;q34)
RUNX1::MECOM, t(3;21)(q26;q22)
RUNX1::RPL22P1, t(3;21)(q26;q22)
KMT2A::AFF1, t(4;11)(q21;q23)
ETV6::PDGFRB, t(5;12)(q33;p13)
NPM1::RARA, t(5;17)(q35;q21)
DEK::NUP214, t(6;9)(p23;q34)
KMT2A::AFDN, t(6;11)(q27;q23)
RUNX1::RUNX1T1, t(8;21)(q22;q22)
SET::NUP214, t(9;9)(q34;q34)
KMT2A::MLLT3, t(9;11)(p21;q23)
ETV6::ABL1, t(9;12)(q34;p13)
BCR::ABL1, t(9;22)(q34;q11)
KMT2A::MLLT10, t(10;11)(p12;q23)
KMT2A PTD, dupMLL(11q23)
ZBTB16::RARA, t(11;17)(q23;q21)
KMT2A::MLLT6, t(11;17)(q23;q21)
KMT2A::ELL, t(11;19)(q23;p13.1)
KMT2A::MLLT1, t(11;19)(q23;p13.3)
ETV6::RUNX1, t(12;21)(p13;q22)
ETV6::MN1, t(12;22)(p13;q12)
PML::RARA, t(15;17)(q24;q21)
CBFB::MYH11, inv(16)(p13;q22)
FUS::ERG, t(16;21)(p11;q22)
TCF3::HLF, t(17;19)(q22;p13)

Bei Anforderungen einzelner Analyten bitte Namen des gewünschten Fusion-Gens auf "Zuweisung zur hämatologischen Diagnostik" bei "Split-out PCR" eintragen

Hämatoonkologie Panel

NGS für myeloische und lymphatische Erkrankungen (letzte Überarbeitung 10.03.2025)

Gene mit vollständiger Abdeckung der kodierenden Sequenz:

BCOR, BCORL1, CEBPA, CREBBP, CSF3R, DDX41, DNMT3A, ETV6, EZH2, GATA2, JAK2, NF1, PHF6, RAD21, RUNX1, SH2B3, STAG2, TET2, TP53, ZRSR2

Gene mit Erfassung von Hotspots:

ABL1, ANKRD26, ASXL1, BIRC3, BRAF, BTK, CALR, CBL, CRLF2, CXCR4, FLT3, HRAS, IDH1, IDH2, IL7RA, JAK1, JAK3, KIT, KRAS, MPL, MYD88, NOTCH1, NPM1, NRAS, PLCG2, PPM1D, PTPN11, SETBP1, SF3B1, SRSF2, STAT3, STAT5B, STAT6, U2AF1, WT1

AmpliSeq Panel

NGS für Onkologie (solide Tumore, z.B. Colon, Lunge, GIST,...), letzte Überarbeitung 10.03.2025

Gene mit Erfassung von Hotspots:

ABL1, AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, FLT3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2, JAK2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53, VHL

TruSight Oncology 500 Panel (TSO500)

NGS für solide Tumore, z.B. Mutationssuche, Fusionen, CNVs, MSI, TMB, optional HRD, letzte Überarbeitung 10.03.2025

A) TSO500 Detektion von Varianten (Punktmutationen, Indels, Amplifikationen)

ABL1, ABL2, ACVR1, ACVR1B, AKT1, AKT2, AKT3, ALK, ALOX12B, ANKRD11, ANKRD26, APC, AR, ARAF, ARFRP1, ARID1A, ARID1B, ARID2, ARID5B, ASXL1, ASXL2, ATM, ATR, ATRX, AURKA, AURKB, AXIN1, AXIN2, AXL, B2M, BAP1, BARD1, BBC3, BCL10, BCL2, BCL2L1, BCL2L11, BCL2L2, BCL6, BCOR, BCORL1, BCR, BIRC3, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRD4, BRIP1, BTG1, BTK, C11orf30, CALR, CARD11, CASP8, CBFB, CBL, CCND1, CCND2, CCND3, CCNE1, CD274, CD276, CD74, CD79A, CD79B, CDC73, CDH1, CDK12, CDK4, CDK6, CDK8, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDKN2C, CEBPA, CENPA, CHD2, CHD4, CHEK1, CHEK2, CIC, CREBBP, CRKL, CRLF2, CSF1R, CSF3R, CSNK1A1, CTCF, CTLA4, CTNNNA1, CTNNB1, CUL3, CUX1, CXCR4, CYLD, DAXX, DCUN1D1, DDR2, DDX41, DHX15, DICER1, DIS3, DNAJB1, DNMT1, DNMT3A, DNMT3B, DOT1L, E2F3, EED, EGFL7, EGFR, EIF1AX, EIF4A2, EIF4E, EML4, EP300, EPCAM, EPHA3, EPHA5, EPHA7, EPHB1, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ERRFI1, ESR1, ETS1, ETV1, ETV4, ETV5, ETV6, EWSR1, EZH2, FAM123B, FAM175A, FAM46C, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAS, FAT1, FBXW7, FGF1, FGF10, FGF14, FGF19, FGF2, FGF23, FGF3, FGF4, FGF5, FGF6, FGF7, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FGFR4, FH, FLCN, FLI1, FLT1, FLT3, FLT4, FOXA1, FOXL2, FOXO1, FOXP1, FRS2, FUBP1, FYN, GABRA6, GATA1, GATA2, GATA3, GATA4, GATA6, GEN1, GID4, GLI1, GNA11, GNA13, GNAQ, GNAS, GPR124, GPS2, GREM1, GRIN2A, GRM3, GSK3B, H3F3A, H3F3B, H3F3C, HGF, HIST1H1C, HIST1H2BD, HIST1H3A, HIST1H3B, HIST1H3C, HIST1H3D, HIST1H3E, HIST1H3F, HIST1H3G, HIST1H3H, HIST1H3I, HIST1H3J, HIST2H3A, HIST2H3C, HIST2H3D, HIST3H3, HLA-A, HLA-B, HLA-C, HNF1A, HNRNP, HOXB13, HRAS, HSD3B1, HSP90AA1, ICOSLG, ID3, IDH1, IDH2, IFNGR1, IGF1, IGF1R, IGF2, IKBKE, IKZF1, IL10, IL7R, INHA, INHBA, INPP4A, INPP4B, IRS1, IRF2, IRF4, IRS1, IRS2, JAK1, JAK2, JAK3, JUN, KAT6A, KDM5A, KDM5C, KDM6A, KDR, KEAP1, KEL, KIF5B, KIT, KLF4, KLHL6, KMT2B, KMT2C, KMT2D, KRAS, LAMP1, LAT51, LAT52, LMO1, LRP1B, LYN, LZTR1, MAGI2, MALT1, MAP2K1, MAP2K2, MAP2K4, MAP3K1, MAP3K13, MAP3K14, MAP3K4, MAPK1, MAPK3, MAX, MCL1, MDC1, MDM2, MDM4, MED12, MEF2B, MEN1, MET, MGA, MITF, MLH1, MLL, MLLT3, MPL, MRE11A, MSH2, MSH3, MSH6, MST1, MST1R,

MTOR, MUTYH, MYB, MYC, MYCL1, MYCN, MYD88, MYOD1, NAB2, NBN, NCOA3, NCOR1, NEGR1, NF1, NF2, NFE2L2, NFKBIA, NKX2-1, NKX3-1, NOTCH1, NOTCH2, NOTCH3, NOTCH4, NPM1, NRAS, NRG1, NSD1, NTRK1, NTRK2, NTRK3, NUP93, NUTM1, PAK1, PAK3, PAK7, PALB2, PARK2, PARP1, PAX3, PAX5, PAX7, PAX8, PBRM1, PDCD1, PDCD1LG2, PDGFRA, PDGFRB, PDK1, PDPK1, PGR, PHF6, PHOX2B, PIK3C2B, PIK3C2G, PIK3C3, PIK3CA, PIK3CB, PIK3CD, PIK3CG, PIK3R1, PIK3R2, PIK3R3, PIM1, PLCG2, PLK2, PMAIP1, PMS1, PMS2, PNRC1, POLD1, POLE, PPARG, PPM1D, PPP2R1A, PPP2R2A, PPP6C, PRDM1, PREX2, PRKAR1A, PRKCI, PRKDC, PRSS8, PTCH1, PTEN, PTPN11, PTPRD, PTPRS, PTPRT, QKI, RAB35, RAC1, RAD21, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RAF1, RANBP2, RARA, RASA1, RB1, RBM10, RECQL4, REL, RET, RFWD2, RHEB, RHOA, RICTOR, RIT1, RNF43, ROS1, RPS6KA4, RPS6KB1, RPS6KB2, RPTOR, RUNX1, RUNX1T1, RYBP, SDHA, SDHAF2, SDHB, SDHC, SDHD, SETBP1, SETD2, SF3B1, SH2B3, SH2D1A, SHQ1, SLT2, SLX4, SMAD2, SMAD3, SMAD4, SMARCA4, SMARCB1, SMARCD1, SMC1A, SMC3, SMO, SNCAIP, SOCS1, SOX10, SOX17, SOX2, SOX9, SPEN, SPOP, SPTA1, SRC, SRSF2, STAG1, STAG2, STAT3, STAT4, STAT5A, STAT5B, STK11, STK40, SUFU, SUZ12, SYK, TAF1, TBX3, TCEB1, TCF3, TCF7L2, TERC, TERT, TET1, TET2, TFE3, TFRC, TGFB1, TGFB2, TMEM127, TMPRSS2, TNFAIP3, TNFRSF14, TOP1, TOP2A, TP53, TP63, TRAF2, TRAF7, TSC1, TSC2, TSHR, U2AF1, VEGFA, VHL, VTCN1, WISP3, WT1, XIAP, XPO1, XRCC2, YAP1, YES1, ZBTB2, ZBTB7A, ZFHX3, ZNF217, ZNF703, ZRSR2

B) TSO500 Detektion von Fusionen

ABL1, AKT3, ALK, AR, AXL, BCL2, BRAF, BRCA1, BRCA2, CDK4, CSF1R, EGFR, EML4, ERBB2, ERG, ESR1, ETS1, ETV1, ETV4, ETV5, EWSR1, FGFR1, FGFR2, FGFR3, FGFR4, FLI1, FLT1, FLT3, JAK2, KDR, KIF5B, KIT, KMT2A, MET, MLLT3, MSH2, MYC, NOTCH1, NOTCH2, NOTCH3, NRG1, NTRK1, NTRK2, NTRK3, PAX3, PAX7, PDGFRA, PDGFRB, PIK3CA, PPARG, RAF1, RET, ROS1, RPS6KB1, TMPRSS2

TruSight Hereditary Cancer Panel

NGS für erbliche Tumorprädispositionssyndrome, letzte Überarbeitung 10.03.2025

ACD, AIP, AKT1, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CHEK2, CTRC, DDB2, DICER1, DIS3L2, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NSD1, NTHL1, PALB2, PDGFRA, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD50, RAD51, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, SPRED1, STK11, SUFU, TERF2IP, TERT, TMEM127, TP53, TSC1, TSC2, VHL, WT1, XPA, XPC, XRCC2

Twist Exome Panel 2.0

Virtuelle Genpanels aus Twist Exome Panel 2.0, letzte Überarbeitung 10.03.2025

Die virtuellen Genpanels enthalten Gene, die laut heutigem Wissensstand mit der jeweiligen Erkrankung assoziiert sind. Abhängig von den klinischen Angaben können im individuellen Fall auch weitere Gene untersucht werden. Die Liste der untersuchten Gene wird am Befund angehängt.

A) Amyloidose

APOA1, APOA2, APOC2, APOC3, B2M, CST3, FGA, GSN, LYZ, NLRP3, TTR

B) Diabetes und MODY

ABCC8, AGPAT2, AGPS, AKT2, ALMS1, APPL1, BLK, BLM, BSCL2, CAV1, CAVIN1, CEL, CIDE, CISD2, DCAF17, DMXL2, DNAJC3, DUT, DYRK1B, EIF2AK3, ENPP1, FGFR3, FOXC2, FOXP3, GATA4, GATA6, GCK, GLIS3, HAMP, HFE, HJV, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, KCNJ11, KLF11, LIPC, LMNA, LRBA, MNX1, MT-TL1, NEUROD1, NEUROG3, NKX2-2, NSMCE2, PAX4, PAX6, PCBD1, PCNT, PCYT1A, PDX1, PIK3R1, PLIN1, POC1A,

POLD1, PPARG, PPP1R15B, PPP1R3A, PSMB8, PTF1A, RFX6, SLC19A2, SLC29A3, SLC2A2, SLC40A1, SMPD4, STAT1, STAT3, TFR2, TRMT10A, WFS1, WRN, ZBTB20, ZFP57, ZMPSTE24

C) Erythrozytose (erbliche Formen)

BPGM, EGLN1/2/3, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, HIF1A, JAK2, PIEZO1, PKLR, SH2B3, SLC30A10, VHL

D) Kardiologische Erkrankungen

- **Hypertrophe Kardiomyopathie (laut EMQN Guidelines 2023, PMID: 37443332)**

ACTC1, FHL1, FLNC, GLA, LAMP2, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKAG2, PTPN11, RAF1, RIT1, TNNT3, TNNT2, TPM1, TTR

- **Brugada Syndrom**

SCN5A, KCNH2

- **Arrhythmogene rechtsventrikuläre Kardiomyopathie (laut EMQN Guidelines 2023, PMID: 37443332)**

DES, DSC2, DSG2, DSP, FLNC, JUP, PKP2, PLN, TMEM43

- **Dilatative Kardiomyopathie (laut EMQN Guidelines 2023, PMID: 37443332)**

ACTC1, ACTN2, BAG3, DES, DMD, DSP, FLNC, JPH2, LMNA, MYH7, NEXN, PLN, RBM20, SCN5A, TNNT1, TNNT3, TNNT2, TPM1, TTN, VCL

- **Linksventrikuläre Non-Compaction Kardiomyopathie (LVNC)**

ACTC1, MYBPC3, MYH7, TNNT2, TPM1

- **Long QT Syndrom (laut EMQN Guidelines 2023, PMID: 37443332)**

CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A, TECRL, TRDN

- **Short QT Syndrom**

CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCN5A, SLC4A3

E) Neuromuskuläre Erkrankungen

AARS1, ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGL, AGRN, AK9, ALDOA, ALG14, ALG2, ALS2, AMACR, AMPD1, ANO5, AR, ASAHI, ASCC1, ASCC3, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, BSCL2, BVES, CACNA1A, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHD8, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ4, COQ8A, COX6A2, CPT1A, CPT1B, CPT2, CRPPA, CRYAB, CYP2C8, DAG1, DCTN1, DES, DGUOK, DHX16, DMD, DMPK, DNA2, DNAJB2, DNAJB4, DNAJB6, DNM2, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DTNA, DUX4L1, DYNC1H1, DYSF, ECEL1, EGR2, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FAM111B, FBP2, FBXO38, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GDAP1, GFER, GFPT1, GGPS1, GIPC1, GLA, GLDC, GLRA1, GLRB, GMPPB, GNE, GOLGA2, GOSR2, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HINT1, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HNRNPU, HRAS, HSPB1, HSPB3, HSPB8, HTRA2, IGHMBP2, INPP5K, ISCU, ITGA7, JAG2, KBTBD13, KIF21A, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LETM1, LGI4, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4, MACF1, MAP3K20, MATR3, MEG3, MEGF10, MFN2, MGME1, MICU1, MLIP, MPZ, MSTO1, MT-CO1, MT-CO2, MTM1, MTMR14, MTO1, MTRFR, MT-TL1, MUSK, MYBPC1, MYBPC3, MYF5, MYF6, MYH1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NEB, NEFL, NTRK1, OBSCN, OPA1,

ORAI1, PABPN1, PAX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG1, PHOX2B, PIEZO2, PLEC, PLEKHG5, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PPA2, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, REEP1, RNASEH1, RPH3A, RRM2B, RXYLT1, RYR1, RYR3, SBDS, SCN4A, SCN9A, SCO2, SELENON, SETX, SGCA, SGCB, SGCD, SGCG, SIGMAR1, SIL1, SLC18A3, SLC22A12, SLC22A5, SLC25A1, SLC25A20, SLC25A26, SLC25A3, SLC25A32, SLC25A4, SLC25A42, SLC2A9, SLC52A1, SLC52A2, SLC52A3, SLC5A7, SLC6A5, SMCHD1, SMN1, SMPX, SNAP25, SNRPN, SOD1, SORD, SPEG, SPG11, SPG7, SPTBN4, SQSTM1, SRPK3, STAC3, STIM1, STIM2, SUCLA2, SVIL, SYNE1, SYNE2, SYT15, SYT2, TAFAZZIN, TANGO2, TCAP, TIA1, TK2, TMEM126B, TMEM43, TNNC2, TNNT2, TNNT1, TNNT3, TNPO3, TNXB, TOR1AIP1, TPM2, TPM3, TRAPP11, TRDN, TRIM32, TRIP4, TRMT5, TRPV4, TSEN54, TSFM, TTN, TUBB3, TWNK, TYMP, UBA1, UBQLN1, UNC13A, UNC45B, VAMP1, VAPB, VCP, VMA21, VPS33B, VRK1, YARS2, ZC4H2

F) Hereditäre Neuropathie

AAAS, AARS1, ABCA1, ABCC9, ABCD1, ABHD12, ACOX1, ACTC1, ACTN2, ADPRS, AFG3L2, AGTPBP1, AGXT, AIFM1, ALDH18A1, ALDH3A2, AMACR, AMPD2, ANKRD1, AP1S1, APOA1, APTX, ARHGEF10, ARL6IP1, ARPC3, ARSA, ASA1, ASCC1, ATAD3A, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BANF1, BCKDHB, BICD2, BRAF, BSCL2, C19orf12, C1orf94, CACNB4, CADM3, CASQ2, CAV3, CCT5, CD59, CFAP276, CHCHD10, CLP1, CLTCL1, CNTNAP1, COA7, COQ7, COQ8A, COX10, COX20, COX6A1, CPOX, CRYAB, CSRP3, CTDP1, CYP27A1, CYP2U1, CYP7B1, DARS2, DCAF8, DCTN1, DDHD1, DEGS1, DES, DGAT2, DGUOK, DHH, DHTKD1, DHX9, DMD, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DSC2, DSG2, DSP, DST, DSTYK, DTNA, DYNC1H1, EGR2, ELP1, EMD, EMILIN1, ERBB3, ERCC6, ERCC8, ETFDH, EXOSC3, EXOSC8, EXOSC9, FA2H, FAH, FAM126A, FBLN5, FBXO38, FDX2, FGD4, FGF14, FICD, FIG4, FKTN, FLVCR1, FXN, GAA, GALC, GAN, GARS1, GATAD1, GBA2, GBE1, GBF1, GDAP1, GJB1, GJB3, GJC2, GLA, GLE1, GNB4, GSN, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, IARS2, IFRD1, IGHMBP2, INF2, IQGAP3, ITPR1, ITPR3, JAG1, JPH2, JUP, KARS1, KCNA1, KCNA2, KCNC3, KIF1A, KIF1B, KIF5A, KLC2, KLHL13, KRAS, L1CAM, LAMA4, LAMP2, LAS1L, LDB3, LITAF, LMNA, LRSAM1, LYST, MAG, MAP1B, MAP2K1, MAP2K2, MARS1, MCM3AP, MED25, MFF, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MRE11, MT-ATP6, MTMR2, MT-ND6, MTRFR, MT-RNR1, MT-TL1, MTTP, MYBPC3, MYH14, MYH6, MYH7, MYL2, MYL3, MYO9B, MYOZ2, MYPN, NAGA, NAGLU, NARS1, NDC1, NDRG1, NEBL, NEFH, NEFL, NEMF, NEXN, NGF, NGLY1, NIPA1, NOTCH2NLA, NRAS, NRG1, NTRK1, NUDT2, OPA1, OPA3, PCK2, PCYT2, PDHA1, PDK3, PDLIM3, PDXK, PDYN, PEX10, PEX12, PEX7, PHYH, PIGB, PKP2, PLA2G6, PLAAT3, PLEKHG5, PLN, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PPOX, PRDM12, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PSMC3, PTEN, PTPN11, PTRH2, RAB7A, RAF1, RBM20, RBM7, REEP1, RETREG1, RFC1, RIT1, RTN2, RYR2, SACS, SARS1, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN5A, SCN9A, SCO2, SCP2, SCYL1, SELENOI, SEPTIN9, SETX, SGCD, SGPL1, SH3BP4, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC1A3, SLC25A19, SLC25A46, SLC52A1, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SMN1, SNAP29, SORD, SOS1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTAN1, SPTBN2, SPTBN4, SPTLC1, SPTLC2, STUB1, SUCLA2, SURF1, SYT2, TAFAZZIN, TCAP, TDP1, TECPR2, TFG, TMEM43, TNNC1, TNNT3, TNNT2, TPM1, TRIM2, TRIP4, TRPA1, TRPV4, TTBK2, TTN, TPPA, TTR, TUBB3, TWNK, TYMP, UBA1, UBA5, UCHL1, VAPPB, VCL, VCP, VPS13A, VRK1, VWA1, WARS1, WASHC5, WNK1, XK, XPA, XRCC1, YARS1, ZFHX3, ZFYVE26, ZFYVE27

G) Nephrolithiasis / Nephrokalzinose

ADCY10, AGK, AGXT, AP2S1, APRT, ATP6VOA4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN14, CLDN16, CLDN19, CYP24A1, FAM20A, FGF23, G6PC1, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, MOCOS, OCRL, PHEX, RRAGD, SLC12A1, SLC17A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC4A1, SLC6A19, SLC6A20, SLC7A9, SLC9A3, SLC9A3R1, STRADA, TRPM6, VDR, VIPAS39, VPS33B, XDH, ZNF365

H) Zystische Nierenerkrankungen

ADAMTS9, AHI1, ALG5, ALG8, ALG9, ALMS1, ANKS6, ARL13B, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CCDC28B, CENPF, CEP104, CEP164, CEP290, CEP41, CEP55, CEP83, CFAP418, CFAP47, CILK1, CLCN5, COL4A1, COL4A3, COL4A4, COL4A5, CPLANE1,

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CRB2, CSPP1, CYP24A1, CYS1, DCDC2, DDX59, DHCR7, DLG5, DNAJB11, DYNC2H1, DYNC2I1, DZIP1L, EXOC3L2, EXOC8, FLCN, GANAB, GLA, GLIS2, HNF1B, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, MUC1, NEK1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PDE6D, PDIA6, PIBF1, PKD1, PKD2, PKHD1, PMM2, PRKCSH, PSKH1, RPGRIP1L, SCLT1, SDCCAG8, SEC61A1, SEC63, SLC41A1, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRIM32, TSC1, TSC2, TTC21B, TTC8, TULP3, TXNDC15, UMOD, VHL, WDPCP, WDR19, WDR35, XPNPEP3, ZNF423

I) Pankreatitis

CASR, CCL2, CEL, CELA3B, CFTR, CLDN2, CPA1, CTRB1, CTRB2, CTRC, CTSB, CXCL8, KRT8, MORC4, PRSS1, SPINK1, TRPV6

J) Cholestase

ABCB4, ABCB11, ABCC2, ADK, AKR1D1, ALDOB, AMACR, AP1S1, ATP7B, ATP8B1, BAAT, BCS1L, CC2D2A, CFTR, CLDN1, COG7, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, FAH, FARSA, FARSB, GALE, GALK1, GALM, GALT, GBA, GBE1, GNAS, HADHA, HNF1B, HSD3B7, IARS1, JAG1, KIF12, LIPA, LSR, MMP15, MPI, MPV17, MVK, MYO5B, NBAS, NOTCH2, NPC1, NPC2, NPHP3, NR1H4, PEX1, PEX12, PEX14, PEX2, PEX26, PEX6, PKHD1, POLG, PPM1F, PSKH1, RINT1, RPGRIP1L, SERPINA1, SLC25A13, SLC51A, SMPD1, TALDO1, TJP2, TMEM67, TRMU, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, VPS50, WDR83OS, YARS1, ZFYVE19

cfDNA DNA Hotspot Panel für solide Tumore (Archer LiquidPlex Solid Tumor Panel, letzte Überarbeitung 10.03.2025, nicht im Akkreditierungsumfang)

AKT1, ALK, AR, BRAF, CDK6, CTNNB1, EGFR, ERBB2, ERBB3, ESR1, FGFR1, FGFR2, FGFR3, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, RET, ROS1, TP53