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Ordens klinikum Linz | Labor für Molekulargenetische Diagnostik

Genlisten Paneldiagnostik

SOP04: Multiplex PCR (Endpunkt-PCR Humangenomanalyse aus isolierten Nukleinsäuren) Ausgabedatum 01.08.2019

KMT2A::FOXO4, t(X;11)(q13;q23)
STIL, SIL::TAL1
KMT2A::MLLT11, t(1;11)(p21;q23)
KMT2A::EPS15, t(1;11)(q32;q23)
TCF3::PBX1, t(1;19)(q23;p13)
NPM1::ALK, t(2;5)(p23;q35)
NPM1::MLF1, t(3;5)(q25;q34)
RUNX1::MECOM, t(3;21)(q26;q22)
RUNX1::RPL22P1, t(3;21)(q26;q22) (Neu ab 25.10.2022)
KMT2A::AFF1, t(4;11)(q21;q23)
ETV6::PDGFR, t(5;12)(q33;p13)
NPM1::RARA, t(5;17)(q35;q22)
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)(p22;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;q11-12) (Neu ab 25.10.2022)
PML::RARA, t(15;17)(q21;q22)
CBFB::MYH11, inv(16)(p13q22)
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 hämato-onkologischen Diagnostik” bei “Split-out PCR” eintragen

SOP18: Hämatoonkologie Panel

NGS für myeloische und lymphatische Erkrankungen, Ausgabedatum 24.01.2023

Gene mit vollständiger Abdeckung der kodierenden Sequenz:
BCOR, CEBPA, CSF3R, CXCR4, DNMT3A, ETV6, EZH2, JAK2, NF1, RUNX1, STAG2, TET2, TP53, ZRSR2

Gene mit Erfassung von Hotspots:
ASXL1, BIRC3, BRAF, BTK, CALR, CBL, FLT3, HRAS, IDH1, IDH2, KIT, KRAS, MPL, MYD88, NOTCH1, NPM1, NRAS, PLCG2, PTPN11, SETBP1, SF3B1, SRSF2, U2AF1, WT1

SOP17: AmpliSeq Panel

NGS für Onkologie (solide Tumore, z.B. Colon, Lunge, GIST,…), Ausgabedatum 24.01.2023

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

SOP14: TruSight Oncology 500 Panel (TSO500)

NGS für solide Tumore, z.B. Mutationssuche, Fusionen, CNVs, MSI, TMB, optional HRD, Ausgabedatum 05.05.2023

A) TSO500 Detektion von Varianten (Punktmutationen und Indels)

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, CTNNA1, 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, HNRNPK, HOXB13, HRAS, HSD3B1, HSP90AA1, ICOSLG, ID3, IDH1, IDH2, IFNGR1, IGF1, IGF1R, IGF2, IKBKE, IKZF1, IL10, IL7R, INHA, INHBA, INPP4A, INPP4B, INSR, IRF2, IRF4, IRS1, IRS2, JAK1, JAK2, JAK3, JUN, KAT6A, KDM5A, KDM5C, KDM6A, KDR, KEAP1, KEL, KIF5B, KIT, KLF4, KLHL6, KMT2B, KMT2C, KMT2D, KRAS, LAMP1, LATS1, LATS2, 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, SLIT2, 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, TGFBR1, TGFBR2, 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

C) TSO 500 Detektion von Amplifikationen

AKT2, ALK, AR, ATM, BRAF, BRCA1, BRCA2, CCND1, CCND3, CCNE1, CDK4, CDK6, CHEK1, CHEK2, EGFR, ERBB2, ERBB3, ERCC1, ERCC2, ESR1, FGF1, FGF10, FGF14, FGF19, FGF2, FGF23, FGF3, FGF4, FGF5, FGF6, FGF7, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FGFR4, JAK2, KIT, KRAS, LAMP1, MDM2, MDM4, MET, MYC, MYCL1, MYCN, NRAS, NRG1, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PTEN, RAF1, RET, RICTOR, RPS6KB1, TFRC

SOP16: TruSight Hereditary Cancer Panel
NGS für erbliche Tumorprädispositionssyndrome, Ausgabedatum 24.01.2023

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

SOP12: Twist Exome Panel 2.0
Virtuelle Genpanels aus Twist Exome Panel 2.0, Ausgabedatum 25.05.2023:

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, AKT2, APPL1, BLK, BSCL2, CEL, CISD2, DCAF17, DNAJC3, DYRK1B, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, KCNJ11, KLF11, LIPC, LMNA, LRBA, MNX1, MT-TL1, NEUROD1, NEUROG3, NKX2-2, PAX4, PAX6, PCBD1, PDX1, PIK3R1, PLIN1, POLD1, PPARG,

PPP1R15B, PTF1A, RFX6, SLC19A2, SLC29A3, SLC2A2, STAT1, STAT3, TRMT10A, WFS1, 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) Glykosylierungserkrankungen (CDG)

ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6AP1, ATP6V0A2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT1, B4GALT7, C1GALT1C1, CAD, CCDC115, CHST14, CHST3, CHST6, CHSY1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EXT1, EXT2, FKRP, FKTN, FUT8, GALNT12, GALNT3, GFPT1, GLS, GMPPB, GNE, GORAB, ISPD, LARGE1, LFNG, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, NUS1, PGAP2, PGAP3, PGM1, PGM3, PIGA, PIGL, PIGM, PIGN, PIGO, PIGS, PIGT, PIGV, PIGW, PMM2, POMGNT1, POMGNT2, POMT1, POMT2, RFT1, RXYLT1, SEC23B, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A8, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TMEM165, TMEM199, TRAPPC11, TUSC3, XYLT1, XYLT2

E) Kardiologische Erkrankungen

- **Hypertrophe Kardiomyopathie**

ACTC1, ACTN2, ALPK3, ATAD3A, CACNA1C, CSRP3, FHL1, FHOD3, FLNC, GLA, GYG1, JPH2, LAMP2, MT-TI, MYBPC3, MYH7, MYL2, MYL3, MYLK2, MYPN, PLN, PRKAG2, PTPN11, RAF1, RIT1, RPS6KB1, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTR, TULP3

- **Brugada Syndrom**

SCN5A, KCNH2

- **Arrhythmogene rechtsventrikuläre Kardiomyopathie**

ANK2, CDH2, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, PLN, TMEM43

- **Dilatative Kardiomyopathie**

ABCC9, ACTC1, ACTN2, BAG3, CSRP3, DES, DMD, DSP, EPG5, EYA4, FKTN, FLNC, HAMP, HFE, HJV, IDH2, JPH2, LDB3, LMNA, MYBPC3, MYH6, MYH7, NEXN, PLN, PPP1R13L, PRDM16, RBM20, SCN5A, SGCD, SLC40A1, SPEG, TAFAZZIN, TCAP, TFR2, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL, WWTR1

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

ACTC1, MYBPC3, MYH7, TNNT2, TPM1, WWTR1

- **Long QT Syndrom**

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

- **Short QT Syndrom**

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

F) Kongenitale Myopathie

ACTA1, ACTN2, ADSS1, AR, ASCC1, ASCC3, BIN1, CACNA1S, CCDC78, CFL2, CNTN1, COL12A1, COL13A1, COL25A1, COL6A1, COL6A2, COL6A3, COX6A2, DHX16, DMPK, DNAJB4, DNM2, DOK7, ECEL1, EPG5, FKBP14, FLNC, FXR1, GBE1, GFER, HACD1, HNRNPA2B1, KBTBD13, KLHL40, KLHL41, KY, LAMP2, LMNA, LMOD3, MAP3K20, MEGF10, MICU1, MLIP, MTM1, MTMR14, MYBPC1, MYBPC3, MYF5, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYO18B, MYOD1, MYPN, NEB, NEFL, ORAI1, PAX7, PIEZO2, PPA2, PYROXD1, RYR1, RYR3, SCN4A, SELENON, SLC25A4, SLC25A42, SPEG, SPTBN4, STAC3, STIM1, SVIL, TNNC2, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRDN, TRIP4, TTN, UNC45B, VMA21, ZC4H2

G) Hereditäre Neuropathie

AARS1, ABCA1, ABHD12, AGTPBP1, AGXT, AIFM1, AP1S1, APOA1, APTX, ARHGEF10, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CD59, CFAP276, CHCHD10, CNTNAP1, COA7, COX6A1, CPOX, CTDP1, CYP27A1, DARS2, DCTN1, DEGS1, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, ETFDH, FAH, FAM126A, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FXN, GALC, GAN, GARS1, GBA2, GDAP1, GJB1, GJC2, GLA, GNB4, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HSPB1, HSPB8, IARS2, IGHMBP2, INF2, JAG1, KCNA2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, LYST, MCM3AP, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTTP, MYH14, NAGA, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, OPA3, PDHA1, PDYN, PEX10, PEX7, PHYH, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, PPOX, PRDM12, PRKCG, PRNP, PRPS1, PRX, PTEN, PTPN11, PTRH2, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN9A, SCYL1, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SORD, SOX10, SPAST, SPG11, SPG7, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TFG, TRIM2, TRPA1, TRPV4, TTPA, TTR, TUBB3, TWNK, TYMP, VCP, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, XRCC1, YARS1, ZFYVE26

H) Nierensuperpanel – schmal

ACE, ACTG2, ACTN4, AGT, AGTR1, AGXT, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6, ANOS1, AP2S1, APOE, APRT, AQP2, ARHGDIA, ARL13B, ARL6, ATP1A1, ATP6V0A4, ATP6V1B1, AVPR2, B9D2, BBS1, BBS10, BBS12, BBS2, BS4, BBS5, BBS7, BBS9, BNC2, BSND, C3, C5orf42, CA2, CASR, CC2D2A, CD151, CD46, CENPF, CEP104, CEP164, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CSPP1, CTNS, CTU2, CUBN, CUL3, CYP24A1, DAAM2, DDX59, DGKE, DHCR7, DLC1, DLG5, DNAJB11, DSTYK, DYNC2H1, DZIP1L, EYA1, FAH, FAM20A, FAM58A, FAT1, FLCN, FN1, FRAS1, FREM1, FREM2, GANAB, GATA3, GATM, GLA, GLI3, GNA11, GON7, GPC3, GREB1L, GRHPR, GRIP1, HAAO, HNF1B, HNF4A, HOGA1, HOXA13, HPRT1, HPSE2, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITSN1, JAG1, KCNJ1, KCNJ10, KCNJ16, KDM6A, KIAA0586, KIAA0753, KIF7, KLHL3, KMT2D, KYNU, LAGE3, LAMB2, LCAT, LIFR, LMX1B, LRIG2, LRP4, LZTFL1, MAGED2, MAGI2, MAPKBP1, MKKS, MKS1, MMACHC, MOCOS, MT-TF, MUC1, MYH9, MYO1E, MYOCD, NADSYN1, NEK8, NIPBL, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C2, NUP107, NUP133, NUP85, NUP93, OCRL, OFD1, OSGEP, PAX2, PBX1, PDSS2, PHEX, PKD1, PKD2, PKHD1, PLCE1, PLVAP, PMM2, PODXL, REN, RET, ROBO2, ROR2, RPGRIP1L, RRAGD, SALL1, SARS2, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SGPL1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A4, SLC5A2, SLC7A9, SMARCAL1, STRA6, STRADA, TBC1D1, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TFAP2A, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM260, TMEM67, TMEM67, TNS2, TP53RK, TPRKB, TRAF3IP1, TRAP1, TRIM8, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, TXNDC15, UMOD, VHL, VIPAS39, VPS33B, WBP11, WDPCP, WDR19, WDR35, WDR60, WDR73, WNK4, WT1, XDH, XPNPEP3, YRDC, ZIC3, ZMYM2

I) Pankreatitis

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

J) Retinopathie

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARSG, ASRGL1, ATF6, ATOH7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP78, CERKL, CFAP410, CFAP418, CFH, CHM, CLCC1, CLN3, CLN5, CLN6, CLN8, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL18A1, COL4A1, COQ2, COQ5, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTSD, CWC27, CYP2R1, CYP4V2, DHDDS, DHX38, DMD, DRAM2, EFEMP1, ELOVL1, ELOVL4, ERCC6, ERCC8, ESPN, EXOSC2, EYS, FAM161A, FLVCR1, FRMD7, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HCCS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT74, IFT81, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, JAG1, KCNJ13, KCNV2, KIAA1549, KIF11, KIF3B, KIZ, KLHL7, LAMA1, LCA5, LIG3, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MAPKAPK3, MED12, MERTK, MFRP, MFSD8, MIR204, MKKS, MKS1, MMACHC, MSTO1, MTRFR, MT-TH, MTTP, MT-TP, MT-TS2, MVK, MYO7A, NBAS, NDP, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPN1LW, OPN1MW, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDSS1, PEX1, PEX2, PEX6, PEX7, PGK1, PHYH, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, POMT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RHO, RIMS2, RLBP1, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SDCCAG8, SEMA4A, SLC24A1, SLC25A46, SLC37A3, SLC38A8, SLC6A6, SNRNP200, SPATA7, SPP2, SPTLC1, SRD5A3, SSBP1, TIMM8A, TIMP3, TINF2, TLCD3B, TMEM216, TMEM218, TMEM231, TMEM237, TOPORS, TPP1, TRAF3IP1, TREX1, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, WDPCP, WDR19, WHRN, ZFYVE26, ZNF408, ZNF423

cfDNA DNA Hotspot Panel für solide Tumore (Archer LiquidPlex Solid Tumor Panel, aktualisiert am 04.04.2023, nicht im Akkreditierungsumfang)

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