

Molekularpathologische Diagnostik

Informationen zur Panelsequenzierung (NGS) – Seite 1 -

nNGM - Beinhaltete molekulare Biomarker:

SNVs: BRAF (Exon 11, 15), EGFR (Exon 18, 19, 20, 21), ALK (Exon 22, 23, 24, 25), CTNNB1 (Exon 3), ERBB2 (HER2, Exons 8, 19, 20), FGFR1 (Exons 4, 5, 6, 7, 10, 12, 13, 14, 15), FGFR2 (Exons 6, 7, 8(b), 9, 10, 11, 12, 13, 14, 15, 18), FGFR3 (Exons 3, 6, 7, 9, 10, 12, 14, 16, 18), FGFR4 (Exons 3, 6, 9, 12, 13, 15, 16), HRAS (Exons 2, 3, 4), IDH1 (Exon 4), IDH2 (Exon 4), KEAP1 (Exon 2, 3, 4, 5, 6), KRAS (Exons 2, 3, 4), MAP2K1 (MEK1, Exons 2, 3), MET (Exons 14, 16, 17, 18, 19 + Intron 13 + 100bp Intron 14), NRAS (Exons 2, 3, 4), NTRK1 (Exons 13, 14, 15, 16, 17), NTRK2 (Exons 14, 15, 16, 17, 18, 19), NTRK3 (Exons 15, 16, 17, 18, 19, 20), PIK3CA (Exons 8, 10, 21), PTEN (Exons 1, 2, 3, 4, 5, 6, 7, 8), RET (Exons 10, 11, 12, 13, 14, 15, 16, 17, 18), ROS1 (Exons 34, 35, 36, 37, 38, 39, 40, 41), STK11 (Exons 1, 2, 3, 4, 5, 6, 7), TP53 (Exons 4, 5, 6, 7, 8)

nNGM inkl. „booster“ - Beinhaltete molekulare Biomarker:

SNVs: BRAF (Exon 11, 15), EGFR (Exon 18, 19, 20, 21), ALK (Exon 22, 23, 24, 25), CTNNB1 (Exon 3), ERBB2 (HER2, Exons 8, 19, 20), FGFR1 (Exons 4, 5, 6, 7, 10, 12, 13, 14, 15), FGFR2 (Exons 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 18), FGFR3 (Exons 3, 6, 7, 9, 10, 12, 14, 16, 18), FGFR4 (Exons 3, 6, 9, 12, 13, 15, 16), HRAS (Exons 2, 3, 4), IDH1 (Exon 4), IDH2 (Exon 4), KEAP1 (Exon 2, 3, 4, 5, 6), KRAS (Exons 2, 3, 4), MAP2K1 (MEK1, Exons 2, 3), MET (Exons 14, 16, 17, 18, 19 + Intron 13 + 100bp Intron 14), NRAS (Exons 2, 3, 4), NTRK1 (Exons 13, 14, 15, 16, 17), NTRK2 (Exons 14, 15, 16, 17, 18, 19), NTRK3 (Exons 15, 16, 17, 18, 19, 20), PIK3CA (Exons 8, 10, 21), PTEN (Exons 1, 2, 3, 4, 5, 6, 7, 8), RET (Exons 10, 11, 12, 13, 14, 15, 16, 17, 18), ROS1 (Exons 34, 35, 36, 37, 38, 39, 40, 41), STK11 (Exons 1, 2, 3, 4, 5, 6, 7), TP53 (Exons 4, 5, 6, 7, 8)

Zusätzlich „Booster“ SNVs: CALR (Exon 9), GNAS (Exon 8, 9), GNAQ (Exon 4, 5), GNA11 (Exon 5), JAK2 (Exon 12, 14), KIT (Exons 9, 11, 13, 17), MPL (Exon 10), PDGFRA (Exons 12, 18)

TruSight Oncology 500 DNA-/RNA-Analytik - Beinhaltete molekulare Biomarker:

DNA - Komplexe Biomarker: Tumor Mutational Burden (TMB); Mikrosatelliten-Stabilität-/Instabilität (MSS/MSI, bzw. MMRp/MMRd)

DNA - Sequenzvarianten: 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, CBFEB, 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, DNMT3L, E2F3, EED, EGFL7, EGFR, EIF1AX, EIF4A2, EIF4E, EML4, EP300, EPCAM, EPHA3, EPHA5, EPHA7, EPHB1, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ERFF1, 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, IKKBE, 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, MAG, MITF, MLH1, MLL, MLLT3, MPL, MRE11A, MSH2, MSH3, MSH6, MST1, MST1R, MTOR, MUTYH, MYB, MYC, MYCL1, MYCN, MYD88, MYO1, 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, PPM1A, PPP2R1A, PPP2R2A, PPP6C, PRDM1, PREX2, PRKAR1A, PRKCI, PRKDC, PRSS8, PTCH1, PTEN, PTPN11, PTPRT, 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, SPOD, 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, ZFXH3, ZNF217, ZNF703, ZRSR2.

DNA - Kopienanzahlveränderungen (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.

RNA - 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, MET, MLL, MLLT3, MSH2, MYC, NOTCH1, NOTCH2, NOTCH3, NRG1, NTRK1, NTRK2, NTRK3, PAX3, PAX7, PDGFRA, PDGFRB, PIK3CA, PPARG, RAF1, RET, ROS1, RPS6KB1, TMPRSS2.

Molekularpathologische Diagnostik

Informationen zur Panelsequenzierung (NGS) – Seite 2 -

Avenio Surveillance - Beinhaltete molekulare Biomarker:

SNVs: ABL1, AKT1, AKT2, ALK, APC, AR, ARAF, BRAF, BRCA1, BRCA2, CCND1, CCND2, CCND3, CD274, CDK4, CDK6, CDKN2A, CSF1R, CTNNB1, DDR2, DPYD, EGFR, ERBB2, ESR1, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, FLT1, FLT3, FLT4, GATA3, GNA11, GNAQ, GNAS, IDH1, IDH2, JAK2, JAK3, KDR, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, MLH1, MSH2, MSH6, MTOR, NF2, NFE2L2, NRAS, NTRK1, PDCG1LG2, PDGFRA, PDGFRB, PIK3CA; PIK3R1, PMS2, PTCH1, PTEN, RAF1, RB1, RET, RNF43, ROS1, SMAD4, SMO, STK11, TERT promoter, TP53, TSC1, TSC2, UGT1A1, VHL

Fusionen: ALK, FGFR2, FGFR3, NTRK1, RET, ROS1

CNV: EGFR, MET

Avenio Expanded - Beinhaltete molekulare Biomarker:

SNVs: ALK, APC, BRAF, BRCA1, BRCA2, DPYD, EGFR, ERBB2, KIT, KRAS, MET, NRAS, PDGFRA, RET, ROS1, TP53, UGT1A1

Fusionen: ALK, RET, ROS1

CNV: EGFR, ERBB2, MET

Weitere Gene: ABCC5, ABCG2, ACTN2, ADAMTS12, ADAMTS16, ARFGEF1, ASTN1, ASTN2, AVPR1A, BCHE, BPIFB4, BRINP2, BRINP3, C6, C6ORF118, CA10, CACNA1E, CDH12, CDH18, CDH8, CDH9, CDKN2A, CHRM2, CNTN5, CNTNAP2, CPXCR1, CPZ, CRMP1, CSMD1, CSMD3, CTNNB1, CTNNB2, CYBB, DCAF12L1, DCAF12L2, DCAF4L2, DCLK1, DCSTAMP, DDI1, DLGAP2, DMD, DNTTIP1, DOCK3, DSC3, DSCAM, EDSCAM, EGFLAM, EPHA6, EYS, FAM135B, FAM151A, FAM71B, FAT1, FBN2, FBXW7, FCRL5, FOXG1, FRYL, GBA3, GBP7, GJA8, GPR139, GRIA2, GRIK3, GRIN2B, GRIN3B, GRM1, GRM5, GRM8, GSX1, HACD1, HCN1, HCRTR2, HEBP1, HECW1, HS3ST4, HS3ST5, HTR1A, HTR1E, IFF16, IL7R, INSL3, ITGA10, ITSN1, KCNA5, KCNB2, KCNC2, KCNJ5; KCTD8, KEAP1, KIAA1211, KIF17, KIF19, KLHL31, KPRP, LPPR4, LRFN5, LRP1B, LRRC7, LRRTM1, LRRTM4, LTBP4, MAP2, MAP7DD3, MKRN3, MMP16, MTX1, MYH7, MYT1L, NAV3, NEUROD4, NFE2L2, NLGN4X, NLRP3, NMUR1, NOL4, NPAP1, NR0B1, NRXN1, NXPH4, NYAP2, OPRD1, P2RY10, PAX6, PCDH15, PDYN, PDZRN3, PGK2, PHACTR1, PIK3CA, PIK3CG, PKHD1L1, POLE, POM121L12, PREX1, RALYL, RFX5, RIN3, RNASE3, ROBO2, SEMA5B, SLC18A3, SLC39A12, SLC6A5, SLC8A1, SLITRK1, SLITRK4, SLITRK5, SLPI, SMAD4, SOX9, SPTA1, ST6GALNAC3, STK11, SV2A, T, THSD7A, TIAM1, TMEM200A, TNFRSF21, TNN, TNR, TRHDE, TRIM58, TRPS1, UGT3A2, USH2A, USP29, VPS13B, WBCSR17, WIPF1, WSCD2, ZC3H12A, ZFPM2, ZIC1, ZIC4, ZNF521, ZSCAN1

TruSight RNA-Fusion - Beinhaltete molekulare Biomarker:

RNA - Fusionen: ABI1, ABL1, ABL2, ACACA, ACE, ACER1, ACKR3, ACSL6, ADD3, AFF1, AFF3, AFF4, AGR3, AHI1, AHRR, ALK, ANKRD28, AR, ARHGAP20, ARHGAP26, ARNT, ASPSCR1, ASTN2, ATF1, ATIC, ATP1B4, AUTS2, BACH2, BAG4, BAIAP2L1, BAZ2A, BCAS3, BCAS4, BCL10, BCL11A, BCL11B, BCL2, BCL2L1, BCL3, BCL6, BCL9, BCOR, BCR, BDNF, BICC1, BIRC3, BIRC6, BRAF, BRD1, BRD3, BRD4, BRWD3, BTBD18, BTG1, C11orf1, C11orf95, C2CD2L, C3orf27, CAMTA1, CAPRIN1, CARS, CASC5, CASP7, CBFA2T3, CBFB, CBL, CCAR2, CCDC28A, CCDC6, CCDC88C, CCNB1IP1, CCNB3, CCND1, CCND2, CCND3, CD74, CDH11, CDK5RAP2, CDK6, CDX1, CDX2, CEBPA, CEBPB, CEPBD, CEBPE, CEP170B, CEP85L, CHD6, CHIC2, CHMP2B, CHST11, CIC, CIITA, CLP1, CLTC, CLTCL1, CMKLR1, CNBP, CNOT2, CNTRL, COG5, COL1A1, COL1A2, COL6A3, COX6C, CPSF6, CRADD, CREB1, CREB3L1, CREB3L2, CREBBP, CRLF2, CRTC1, CSF1, CSF1R, CTDSF2, CTNNB1, CUX1, DAB2IP, DACH1, DACH2, DDI3, DDX10, DDX20, DEK, DMRT1, DNAJB1, DPM1, DUSP22, DUX4, EBF1, EEFSEC, EGFR, EGR1, EGR2, GR3, EGR4, EIF4A2, ELF4, ELK4, ELL, ELN, EML1, EML4, EP300, EP400, EPC1, EPOR, EPS15, ERBB3, ERC1, ERCC1, ERG, ERLIN2, ESR1, ETS1, ETV1, ETV4, ETV5, ETV6, EWSR1, EZR, FAM19A2, FCGR2B, FCRL4, FEN1, FEV, FGF8, FGFR1, FGFR10P, FGFR10P2, FGFR2, FGFR3, FGFR4, FHIT, FIP1L1, FLI1, FLNA, FLT3, FLT3LG, FNBP1, FOSB, FOSL1, FOXO1, FOXO4, FOXP1, FRK, FRYL, FUS, GAS5, GAS7, GATA1, GIT2, GLI1, GOSR1, GOT1, GPR128, GPR34, GRHRP, GRID1, GTF2I, H2AFX, HAS2, HEY1, HEX, HIP1, HIPK1, HIST1H4I, HLF, HMGA2, HNF1A, HOXA10, HOXA11, HOXA13, HOXA9, HOXC11, HOXC13, HOXD11, HOXD13, HSP90AA1, ID4, IKZF1, IL2, IL21R, IL3, INPP5D, IQCG, IRF2BP2, IRF4, IRS4, ITK, JAK1, JAK2, JAZF1, KANK1, KAT6A, KAT6B, KDM5A, KIAA1524, KIF5B, KMT2A, KPNB1, KSR1, LASP1, LCK, LCP1, LGR5, LHFP, LHX2, LHX4, LINC00598, LINC00982, LMBRD1, LMO1, LMO2, LNP1, LPP, LPXN, LRMP, LRRC37B, LTBP1, LYL1, MACROD1, MAF, MAFB, MALT1, MAML2, MAPRE1, MBNL1, MBTD1, MDS2, MEAF6, MECOM, MGEA5, MKL1, MKL2, MLF1, MLLT1, MLLT10, MLLT11, MLLT3, MLLT4, MLLT6, MN1, MNX1, MSI2, MSN, MUC1, MUTYH, MYB, MYBL1, MYC, MYH11, MYH9, MYO18A, MYO1F, NAB2, NAPA, NBEAP1, NBR1, NCOA1, NCOA2, NCOA3, NDE1, NF1, NFATC2, NFIB, NGF, NGFR, NIN, NIPBL, NKX2-5, NONO, NOTCH1, NPM1, NR4A3, NR6A1, NSD1, NT5C2, NTF3, NTF4, NTRK1, NTRK2, NTRK3, NUMA1, NUP107, NUP214, NUP98, NUTM1, NUTM2A, NUTM2B, OFD1, OLIG2, OLR1, OMD, P2RY8, PAPP, PATZ1, PAX3, PAX5, PAX7, PAX8, PBX1, PCM1, PDE4DIP, PDGFB, PDGFRA, PDGFRB, PER1, PHF1, PHF23, PICALM, PIM1, PLAG1, PML, POM121, POU2AF1, POU5F1, PPAP2B, PPARG, PPARGC1A, PPFIBP1, PPP2R1B, PRCC, PRDM16, PRKACA, PRKAR1A, PRKG2, PRRX2, PSIP1, PSMD2, PTPRR, PVT1, RABEP1, RAD51B, RAF1, RANBP2, RAP1GDS1, RARA, RBM15, RBM6, RCOR1, RCSD1, RET, RHOH, RNF213, ROS1, RPL22, RPN1, RREB1, RRM1, RTE1, RUNX1, RUNX1T1, SARNP, SEC31A, SEPT2, SEPT5, SEPT6, SEPT9, SERPINE1, SERPINF1, SET, SETBP1, SFPQ, SH3D19, SH3GL1, SIK3, SLC34A2, SLC45A3, SLC01B3, SMAP1, SMARCA5, SMARCB1, SNHG5, SORBS2, SORT1, SP3, SPECC1, SPTBN1, SQSTM1, SRF, SRSF3, SS18, SS18L1, SSBP2, SSS1, SSS2, SSS4, ST6GAL1, STAT5B, STAT6, STRN, SUGP2, SUZ12, SYK, TACC1, TACC2, TACC3, TAF15, TAL1, TAL2, TAOK1, TBX15, TCF12, TCF3, TCL1A, TCTA, TEAD1, TEAD2, TEAD3, TEAD4, TEC, TENM1, TET1, TFE3, TFG, TFPT, TFR3, TGFB3, THADA, THRAP3, TIRAP, TLX1, TLX3, TMRSS2, TNFRSF17, TOP1, TOP2B, TP53BP1, TPM3, TPM4, TRHDE, TRIM24, TRIP11, TRPS1, USP16, USP42, USP6, VGLL3, WASF2, WDR18, WDR70, WHSC1, WHSC1L1, WSB1, WT1, WWTR1, XIAP, YAP1, YTHDF2, YWHAE, ZBTB16, ZC3H7A, ZC3H7B, ZFP64, ZFPM2, ZFYVE19, ZMIZ1, ZMYM2, ZMYND11, ZNF207, ZNF384, ZNF444, ZNF521, ZNF585B, ZNF687