

## Molekularpathologische Diagnostik

### Informationen zur Panelsequenzierung (NGS)

#### nNGM v3.1 - Beinhaltete molekulare Biomarker:

**DNA-Sequenzvarianten:** *BRAF* (Exon 11, 12, 15, 14), *EGFR* (Exon 18, 19, 20, 21), *ALK* (Exon 20 (teilweise), 21, 22, 23, 24, 25, 26, 27, 28), *CTNNB1* (Exon 3), *CUL3* (Exon 1-17), *ERBB2/HER2* (Exon 8, 18, 19, 20, 21), *FGFR1* (Exon 4, 5, 6, 7, 10, 12, 13, 14, 15, 16), *FGFR2* (Exon 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 18), *FGFR3* (Exon 3, 6, 7, 9, 10, 12, 13, 14, 15, 16, 18), *FGFR4* (Exon 3, 6, 9, 12, 13, 15, 16), *HRAS* (Exon 2, 3, 4), *IDH1* (Exon 4), *IDH2* (Exon 4), *KEAP1* (Exon 2, 3, 4, 5, 6), *KRAS* (Exon 2, 3, 4), *MAP2K1/MEK1* (Exon 2, 3), *MET* (Exon 14, 16, 17, 18, 19 + Intron 13 + 100bp Intron 14), *NFE2L2* (Exon 1, 2, 3, 4, 5), *NRAS* (Exon 2, 3, 4), *NTRK1* (Exon 13, 14, 15, 16, 17), *NTRK2* (Exon 14, 15, 16, 17, 18, 19), *NTRK3* (Exon 15, 16, 17, 18, 19, 20), *PIK3CA* (Exon 8, 10, 21), *PTEN* (Exon 1, 2, 3, 4, 5, 6, 7, 8, 9), *RET* (Exon 10, 11, 12, 13, 14, 15, 16, 17, 18), *ROS1* (Exon 34, 35, 36, 37, 38, 39, 40, 41), *STK11* (Exon 1, 2, 3, 4, 5, 6, 7), *TP53* (Exon 2, 3, 4, 5, 6, 7, 8, 9, 10, 11)

#### nNGM v3.2 - Beinhaltete molekulare Biomarker:

**DNA-Sequenzvarianten:** *BRAF* (Exon 11, 12, 15, 14), *EGFR* (Exon 18, 19, 20, 21), *ALK* (Exon 20 (teilweise), 21, 22, 23, 24, 25, 26, 27, 28), *CTNNB1* (Exon 3), *CUL3* (Exon 1-17), *ERBB2/HER2* (Exon 8, 18, 19, 20, 21), *FGFR1* (Exon 4, 5, 6, 7, 10, 12, 13, 14, 15, 16), *FGFR2* (Exon 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 18), *FGFR3* (Exon 3, 6, 7, 9, 10, 12, 13, 14, 15, 16, 18), *FGFR4* (Exon 3, 6, 9, 12, 13, 15, 16), *HRAS* (Exon 2, 3, 4), *IDH1* (Exon 4), *IDH2* (Exon 4), *KEAP1* (Exon 2, 3, 4, 5, 6), *KRAS* (Exon 2, 3, 4), *MAP2K1/MEK1* (Exon 2, 3), *MET* (Exon 14, 16, 17, 18, 19 + Intron 13 + 100bp Intron 14), *NFE2L2* (Exon 1, 2, 3, 4, 5), *NRAS* (Exon 2, 3, 4), *NTRK1* (Exon 13, 14, 15, 16, 17), *NTRK2* (Exon 14, 15, 16, 17, 18, 19), *NTRK3* (Exon 15, 16, 17, 18, 19, 20), *PIK3CA* (Exon 8, 10, 21), *PTEN* (Exon 1, 2, 3, 4, 5, 6, 7, 8, 9), *RET* (Exon 10, 11, 12, 13, 14, 15, 16, 17, 18), *ROS1* (Exon 34, 35, 36, 37, 38, 39, 40, 41), *STK11* (Exon 1, 2, 3, 4, 5, 6, 7), *TP53* (Exon 2, 3, 4, 5, 6, 7, 8, 9, 10, 11)  
**+ *RB1* (Exon 1-27), *SMARCA4* (Exon 2-36)**

#### nNGM v3.1 inkl. "Booster v3"- Beinhaltete molekulare Biomarker:

**DNA-Sequenzvarianten:** *BRAF* (Exon 11, 12, 15, 14), *EGFR* (Exon 18, 19, 20, 21), *ALK* (Exon 20 (teilweise), 21, 22, 23, 24, 25, 26, 27, 28), *CTNNB1* (Exon 3), *CUL3* (Exon 1-17), *ERBB2/HER2* (Exon 8, 18, 19, 20, 21), *FGFR1* (Exon 4, 5, 6, 7, 10, 12, 13, 14, 15, 16), *FGFR2* (Exon 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 18), *FGFR3* (Exon 3, 6, 7, 9, 10, 12, 13, 14, 15, 16, 18), *FGFR4* (Exon 3, 6, 9, 12, 13, 15, 16), *HRAS* (Exon 2, 3, 4), *IDH1* (Exon 4), *IDH2* (Exon 4), *KEAP1* (Exon 2, 3, 4, 5, 6), *KRAS* (Exon 2, 3, 4), *MAP2K1/MEK1* (Exon 2, 3), *MET* (Exon 14, 16, 17, 18, 19 + Intron 13 + 100bp Intron 14), *NFE2L2* (Exon 1, 2, 3, 4, 5), *NRAS* (Exon 2, 3, 4), *NTRK1* (Exon 13, 14, 15, 16, 17), *NTRK2* (Exon 14, 15, 16, 17, 18, 19), *NTRK3* (Exon 15, 16, 17, 18, 19, 20), *PIK3CA* (Exon 8, 10, 21), *PTEN* (Exon 1, 2, 3, 4, 5, 6, 7, 8, 9), *RET* (Exon 10, 11, 12, 13, 14, 15, 16, 17, 18), *ROS1* (Exon 34, 35, 36, 37, 38, 39, 40, 41), *STK11* (Exon 1, 2, 3, 4, 5, 6, 7), *TP53* (Exon 2, 3, 4, 5, 6, 7, 8, 9, 10, 11)  
**+ „Boostergene“:** *CALR* (Exon 9), *CXCR4* (Exon 2), *ESR1* (Exon 4, 5, 6, 7, 8), *GNAS* (Exon 8, 9), *GNAQ* (Exon 4, 5), *GNA11* (Exon 5), *JAK2* (Exon 12, 14), *KIT* (Exon 9, 11, 13, 17), *MPL* (Exon 10), *MYD88* (Exon 5), *PDGFRA* (Exon 12, 18), *POLD1* (Exon 6, 7, 8, 9, 10, 11, 12), *POLE* (Exon 9, 11, 13, 14), *SF3B1* (Exon 14, 15, 16)

#### QIaseq Targeted RNAscan Fusion - Beinhaltete molekulare Biomarker:

**RNA-Fusionen:** *ALK*, *BAG4*, *BRAF*, *CCDC6*, *CD74*, *CIAO1*, *COPA*, *CUX1*, *EGFR*, *EML4*, *ETV6*, *EZR*, *FGFR1*, *FGFR2*, *FGFR3*, *FGFR4*, *GOPC*, *HIP1*, *KIF5B*, *KLC1*, *LRIG3*, *MET*, *MPRIP*, *MRPS14*, *NRG1*, *NTRK1*, *NTRK2*, *NTRK3*, *RAD51*, *RET*, *ROS1*, *SDC4*, *SLC34A2*, *STRN*, *TACC3*, *TFG*, *TPM3*, *TPR*, *TRIM33*, *UBE3C*

#### Oncomine Precision Assay w3.4.2 Liquid Biopsy cfTNA - Beinhaltete molekulare Biomarker:

**DNA-Sequenzvarianten – nur Hotspot-Bereiche:** *AKT1*, *AKT2*, *AKT3*, *ALK*, *AR*, *ARAF*, *BRAF*, *CDK4*, *CDKN2A*, *CHEK2*, *CTNNB1*, *EGFR*, *ERBB2*, *ERBB3*, *ERBB4*, *ESR1*, *FGFR1*, *FGFR2*, *FGFR3*, *FGFR4*, *FLT3*, *GNA11*, *GNAQ*, *GNAS*, *HRAS*, *IDH1*, *IDH2*, *KIT*, *KRAS*, *MAP2K1*, *MAP2K2*, *MET*, *MTOR*, *NRAS*, *NTRK1*, *NTRK2*, *NTRK3*, *PDGFRA*, *PIK3CA*, *PTEN*, *RAF1*, *RET*, *ROS1*, *SMO*, *TP53*

**Kopienzahlveränderungen:** *ALK*, *AR*, *CD274*, *CDKN2A*, *EGFR*, *ERBB2*, *ERBB3*, *FGFR1*, *FGFR2*, *FGFR3*, *KRAS*, *MET*, *PIK3CA*, *PTEN*

**Fusionen:** *ALK*, *BRAF*, *ESR1*, *FGFR1*, *FGFR2*, *FGFR3*, *MET*, *NRG1*, *NTRK1*, *NTRK2*, *NTRK3*, *NUTM1*, *RET*, *ROS1*, *RSPO2*, *RSPO3*

#### Oncomine Myeloid Assay Gx v2 - Beinhaltete molekulare Biomarker:

**DNA-Sequenzvarianten (gesamtes Gen abgedeckt = unterstrichen):** *ABL1*, *ANKRD26*, *ASXL1*, *BCOR*, *BRAF*, *CALR*, *CBL*, *CEBPA*, *CSF3R*, *DDX41*, *DNMT3A*, *ETV6*, *EZH2*, *FLT3*, *GATA2*, *HRAS*, *IDH1*, *IDH2*, *IKZF1*, *JAK2*, *KIT*, *KRAS*, *MPL*, *MYD88*, *NEF1*, *NPM1*, *NRAS*, *PHE6*, *PPM1D*, *PRPF8*, *PTPN11*, *RB1*, *RUNX1*, *SETBP1*, *SF3B1*, *SH2B3*, *SMC1A*, *SMC3*, *SRSF2*, *STAG2*, *TET2*, *TP53*, *U2AF1*, *WT1*, *ZRSR2*

**RNA-Fusionen:** *ABL1*, *ABL2*, *BCL2*, *BRAF*, *CCND1*, *CREBBP*, *EGFR*, *ETV6*, *FGFR1*, *FGFR2*, *FUS*, *HMGA2*, *JAK2*, *KAT6A* (MOZ), *KAT6B*, *KMT2A*, *MECOM*, *MET*, *MLL10*, *MLL3*, *MRTFA* (MKL1), *MYBL1*, *MYH11*, *NTRK2*, *NTRK3*, *NUP98*, *NUP214*, *PAX5*, *PDGFRA*, *PDGFRB*, *PARA*, *RUNX1*, *TCF3*, *TFE3*, *ZNF384*

