# Untersuchungskatalog Molekularpathologie, Institut für Pathologie und Ambulanzzentrum der MHH

## 1. Klonalitätsanalyse in hämatologischen Neoplasie

| | | | |
|---|---|---|---|
| IGH (Fr1, Fr2, Fr3) | d | Kapillarelektroph. | AA_MP_NA_013_IGH_Klonalitaet |
| IGkappa | d | Kapillarelektroph. | AA_MP_NA_014_IGkappa_Klonalitaet |
| TCRβ, γ, δ | d | Kapillarelektroph. | AA_MP_NA_015_TCRB_Klonalitaet |
| | | | AA_MP_NA_016_TCRD_Klonalitaet |
| | | | AA_MP_NA_017_TCRG_Klonalitaet |
| IGHV-Mutationsst. | pro | PAA-Gel, Sanger-Seq. | AA_MP_NA_002_IGHV_Mutationsstatus |

## 2. Mutationsanalyse einzelner Codons bzw. Exons

| | | | |
|---|---|---|---|
| BRAF (600) | d, pro, prä | Pyroseq., ggf. dPCR | AA_MP_NA_024_Pyro_BRAF_Codon_600 |
| | | | AA_MP_NA_069_dPCR_BRAF_600 |
| BRAF Ex. 11 + 15 | pro, prä | NGS | AA_MP_NA_036_NGS_BRAF_Exon_11_15 |
| CALR Ex. 9 | d | NGS | AA_MP_NA_044_NGS_CALR |
| CBL Ex. 8, 9 | d | NGS | AA_MP_NA_045_NGS_CBL |
| CSF3R Ex. 14, 17 | d | NGS | AA_MP_NA_047_NGS_CSF3R |
| CTNNB1 Ex. 3 | d | NGS | AA_MP_NA_037_NGS_CTNNB1_Exon_3 |
| EGFR Ex. 18-21 | prä | NGS | AA_MP_NA_038_NGS_EGFR_Exon_18_bis_21 |
| ERBB2/HER2-Mut | pro, prä | NGS | AA_MP_NA_032_NGS_ERBB2_HER2 |
| ESR1-Mut | pro, prä | NGS | AA_MP_NA_031_NGS_ESR1 |
| FLT3 Ex. 14, 15, 20 | d, pro | NGS | AA_MP_NA_056_NGS_FLT3 |
| FOXL2 (134) | d | Pyroseq. | AA_MP_NA_025_Pyro_FOXL2_Codon_134 |
| GNAS (201/220) | d | Pyroseq. | AA_MP_NA_026_Pyro_GNAS |
| H3F3A (27/34) | d | Pyroseq. | AA_MP_NA_027_Pyro_H3F3A |
| HIST1H3B (27) | d | NGS | AA_MP_NA_057_NGS_HIST1H3B_Codon_27 |
| HRAS (12/13) | d, prä | Pyroseq. | AA_MP_NA_028_Pyro_HRAS |
| IDH1 (131/132) | d, prä | Pyroseq. | AA_MP_NA_029_Pyro_IDH_1_2 |
| IDH2 (140, 172) | d, prä | Pyroseq. | AA_MP_NA_029_Pyro_IDH_1_2 |
| JAK-2 (617) | d | Pyroseq. | AA_MP_NA_021_Pyro_JAK2_Codon _617 |
| JAK-2 Ex. 12 | d | Pyroseq., ggf. Sanger-Seq. | AA_MP_NA_022_Pyro_JAK2_Exon _12 |



| Ersteller: | Prüfer: | Genehmiger: |
|---|---|---|
| Schipper, Elisa | 13.09.2022, Bartels, Stephan Dr. | 13.09.2022, Lehmann-Mühlenhoff, Ulrich Prof. Dr. |

| | | | |
|---|---|---|---|
| KIT (816) | d, prä | Pyroseq. | AA_MP_NA_082_Pyro_KIT_cDNA |
| KIT Ex. 9, 11, 13, 14, 17 | d, pro, prä | NGS | AA_MP_NA_039_NGS_KIT |
| KRAS (12/13) | d, pro, prä | NGS | AA_MP_NA_058_NGS_KRAS_Codon_12_13 |
| MAP2K1 Ex.2, 3, 7 | d, pro, prä | NGS . | AA_MP_NA_040_NGS_MAP2K1 |
| MED12 Ex. 2 | d | Sanger-Seq. | AA_MP_NA_059_Sanger_MED_12 |
| MPL (515) | d | NGS | AA_MP_NA_055_NGS_MPL |
| MYD88 (265) | d | Pyroseq. | AA_MP_NA_020_Pyro_MYD88 |
| NPM1 Ex. 12 | d, pro | NGS | AA_MP_NA_041_NGS_NPM1_Exon_12 |
| NRAS (12/13) | d | NGS | AA_MP_NA_064_NGS_NRAS_Codon_12_13 |
| NRAS (61) | d, prä | NGS | AA_MP_NA_065_NGS_NRAS_Codon_61 |
| PDGFRa Ex. 12, 14, 18 | d, pro, prä | NGS | AA_MP_NA_042_NGS_PDGFRa |
| PIK3CA Ex. 9 + 20 | d, prä | NGS | AA_MP_NA_033_NGS_PIK3CA |
| RAS erw. | pro, prä | NGS | AA_MP_NA_043_NGS_RAS_erweitert |
| RET (918) | d | NGS | AA_MP_NA_072_NGS_RET_Codon_918 |
| RhoA (17) | d | NGS | AA_MP_NA_073_NGS_RHOA_Codon_17 |
| SETBP1 Ex. 4 | d | NGS | AA_MP_NA_051_NGS_SETBP1_Exon_4 |
| SF3B1 Ex. 14 + 15 | d | NGS | AA_MP_NA_052_NGS_SF3B1 |
| SRSF2 (95) | d | NGS | AA_MP_NA_053_NGS_SRSF2 |
| STAT3 Ex. 21 | d | Sanger-Seq. | AA_MP_NA_007_Sanger_STAT3 |
| TERT-Promotor | d, pro | NGS | AA_MP_NA_035_NGS_TERT_Promotor |
| U2AF1 (34 + 157) | d | NGS | AA_MP_NA_054_NGS_U2AF1 |

## 3. Mutationsanalyse kompletter Gene bzw. umfangreicher Genpanel

| | | | |
|---|---|---|---|
| Basis-Panel | d, pro, prä | NGS | AA_MP_NA_046_NGS_Basis_Panel |
| BRCA1 und 2 | prä | NGS | AA_MP_NA_048_NGS_BRCA |
| CDH1 | d | NGS . | AA_MP_NA_080_NGS_CDH1 |
| CompCancer-DNA | d, pro, prä | NGS | AA_MP_NA_062_NGS_Comprehensive_DNA |
| CompCancer-DNA-plus | d, pro, prä | NGS | AA_MP_NA_001_NGS_Comprehensive_DNA_plus |
| Focus-DNA | d, pro, prä | NGS | AA_MP_NA_063_NGS_Focus_DNA |
| Mamma-Metastasen | d, pro, prä | NGS | AA_MP_NA_050_NGS_Mamma_Metastasen |
| MDS/MPN-Panel_v2 | d, pro, prä | NGS | AA_MP_NA_030_NGS_MDS_MPN_v2 |
| Myeloid-NGS-DNA | d, pro, prä | NGS | AA_MP_NA_060_NGS_Myeloid_DNA |

| | | | | |
|---|---|---|---|---|
| POLE | d, pro | NGS | . | AA_MP_NA_061_NGS_POLE |
| Resistenz-Panel | prä | NGS | . | AA_MP_NA_081_NGS_Resistenz_Panel |
| TP53 | d, pro | NGS | . | AA_MP_NA_034_NGS_TP53 |

## 4. Nachweis von Fusionstranskripten und aberranter RNA-Expression

| | | | |
|---|---|---|---|
| Archer Lung-Fusion | d, prä | NGS | AA_MP_NA_049_NGS_Archer_LungFusion |
| Archer Pan-Solid-Tumor | d, prä | NGS | AA_MP_NA_0086_NGS_Archer_Pan_Solid_Tumor |
| bcr/abl | d, prä | PAA-Gel | AA_MP_NA_012_bcr_abl |
| CompCancer-RNA | prä | NGS | A_MP_NA_067_NGS_Comprehensive_RNA |
| cyclinD1 | d | quant. RT-PCR | AA_MP_NA_078_TaqMan_cyclin_D1 |
| FIP1L1/PDGFRa | d | NGS | AA_MP_NA_068_NGS_FIP1L1_PDGFRa |
| kappa/lambda | d | quant. RT-PCR | AA_MP_NA_077_TaqMan_kappa_lambda |
| KIAA1549-BRAF | d | NGS | AA_MP_NA_003_NGS_KIAA1549_BRAF_Fusion |
| MET Ex 14 skipping | prä | NGS | AA_MP_NE_004_MET_Exon_14_skipping |
| Myeloid-NGS-RNA | d, pro, prä | NGS | AA_MP_NA_066_NGS_Myeloid_RNA |
| NTRK-Fusionen | prä | NGS | AA_MP_NA_005_NGS_NTRK_Fusion |
| RET-Fusionen | d, prä | NGS | AA_MP_NA_006_NGS_RET_Fusion |

## 5. Mutationsanalyse im Blut („liquid biopsy“)

| | | | |
|---|---|---|---|
| BRAF V600E | prä | dPCR | AA_MP_NA_069_dPCR_BRAF_600 |
| EGFR T790M | prä | dPCR | AA_MP_NA_070_dPCR_EGFR_T790M |
| Mamma-LB-Panel | d, prä | NGS | AA_MP_NA_083_NGS_Mamma_LB |
| Basis-NGS-LB | d, pro, prä | NGS | AA_MP_NA_084_NGS_Basis_LB |

## 6. Pathogennachweis

| | | | |
|---|---|---|---|
| Mykobakterien | d | PAA-Gel<br>ggf. Sanger-Seq. | AA_MP_NA_023_Mykobakterien |
| M.tbc-Komplex-Diff. | d | PAA-Gel, ggf. Sanger-Seq. | AA_MP_NA_008_M_tbc_Komplex_Subdifferenzierung |
| HPV | d, pro | Hybrid., ggf. Sanger-Seq. | AA_MP_NA_018_HPV |
| Pilze | d | Hybrid., ggf. Sanger-Seq. | AA_MP_NA_019_Pilze |

## 7. weitere Untersuchungen

| | | | |
|---|---|---|---|
| CDKN2A-Deletion | d, pro | dPCR | AA_MP_NA_071_dPCR_CDKN2A_Deletion |
| Homologe Rekombinationsdefizienz (HRD) | prä | OncoScan SNP-Array | AA_MP_NA_085_OncoScan_HRD |
| MGMT-Methylierung | prä | Pyroseq. | AA_MP_NA_010_Methylierung_MGMT |
| Mikrosatelliten-Instabilität | d, pro, prä | Kapillarelektroph. | AA_MP_NA_079_Fragmentanalyse_MSI |
| MLH1-Methylierung | d | Pyroseq. | AA_MP_NA_011_Methylierung_MLH1 |
| STR-Analyse | d | Kapillarelektroph. | AA_MP_NA_075_Fragmentanalyse_STR |
| Tumormutationslast (TMB) | prä | NGS | AA_MP_NA_076_NGS_TMB |

## Abkürzungen in diesem Dokument

| | |
|---|---|
| d: | diagnostischer Marker |
| pro: | prognostischer Marker |
| prä: | prädiktiver Marker |
| Kapillarelektroph.: | Kapillarelektrophorese |
| Sanger-Seq.: | Sanger-Sequenzierung |
| Pyroseq.: | Pyrosequenzierung |
| NGS: | Next Generation Sequencing |
| dPCR: | digitale PCR |
| quant. RT-PCR | quantitative Reverse-Transkriptase-PCR |
| Hybrid.: | Hybridisierung |
| PAA-Gel: | Polyacrylamid-Gel |

## Mitgeltende Dokumente:

Alle in der rechten Spalte aufgeführten Arbeitsanweisungen (AA).



| Ersteller: | Prüfer: | Genehmiger: |
|---|---|---|
| Schipper, Elisa | 13.09.2022, Bartels, Stephan Dr. | 13.09.2022, Lehmann-Mühlenhoff, Ulrich Prof. Dr. |