

Molekularpathologische Untersuchungen

Patientin/Patient Name: Geb.-Datum:	Einsendende(r) Ärztin/Arzt
--	-----------------------------------

Panel-Diagnostik

Parallele Sequenzierung und Detektion von Single Nucleotide Variations (SNVs), Insertionen/Deletionen, Copy Number Variations (CNVs) und Translokationen/Genfusionen (Genlisten siehe Anhang)

- 52 Gene
- 161 Gene
- 324 Gene
- BRCA1/2 bzw. Homologe Rekombinationsdefizienz - Testung (HRD-LOH Score)

Diagnostik einzelner Gene

Bitte gewünschte Gene eintragen

- _____

Diagnostik weiterer Biomarker

- Mikrosatelliteninstabilität (IHC, PCR, NGS)
- EGFR T790M – Resistenzmutation von Liquid Biopsy/Blut
- MGMT - Methylierung
- MLH1 - Methylierung
- PD-L1 - Expression (TPS, CPS, ICS)
- Tumor Mutational Burden (TMB)

Erreger Diagnostik

- Borrelia burgdorferi*
- Chlamydia trachomatis*
- Helicobacter pylori* Clarithromycin-Resistenz
- HPV High Risk & HPV Low Risk (Subtypisierung)
- Leishmania spp.*
- M. tuberculosis*-Komplex & atypische Mykobakterien
- Orthopox & Monkeypox Viren
- Tropheryma whipplei*

Untersuchungsmaterial

Natives oder Formalin-fixiertes und Paraffin-eingebettetes Gewebe, zytologisches Material, Liquid Biopsy (spezielle Blutentnahmeröhrchen erforderlich; Zusendung auf Anfrage)

Panel-Diagnostik - Genlisten

Detektion von single nucleotide variations, Deletionen/Insertionen, copy number variations und *Translokationen* (in kursiv)

52 Gene

AKT1, ALK, AR, BRAF, CCND1, CDK4, CDK6, CTNNB1, DDR2, EGFR, ERBB2-4, ESR1, FGFR1-4, GNA11, GNAQ, HRAS, IDH1-2, JAK1-3, KIT, KRAS, MAP2K1-2, MET, MTOR, MYC, MYCN, NRAS, PDGFRA, PIK3CA, RAF1, RET, ROS1, SMO

ABL1, ALK, AKT3, AXL, BRAF, EGFR, ERBB2, ERG, ETV1, ETV4, ETV5, FGFR1-3, MET, NTRK1-3, PDGFRA, PPARG, RAF1, RET, ROS1

161 Gene

AKT1-3, ALK, AR, ARAF, ARID1A, ATM, ATR, ATRX, AXL, BAP1, BRAF, BRCA1, BRCA2, BTK, CBL, CCND1-3, CDK2/4/6, CDK12, CDKN1B, CDKN2A/B, CHEK1, CHEK2, CREBBP, CSF1R, CTNNB1, DDR2, EGFR, ERBB2-4, ERCC2, ESR1, EZH2, FANCA, FANCD2, FANCI, FBXW7, FGF19, FGF3, FGFR1-4, FLT3, FOXL2, GATA2, GNA11, GNAQ, GNAS, H3F3A, HIST1H3B, HNF1A, HRAS, IDH1-2, IGF1R, JAK1-3, KDR, KIT, KNSTRN, KRAS, MAGOH, MAP2K1/2/4, MAPK1, MAX, MDM2/4, MET, MLH1, MRE11A, MSH2, MSH6, MTOR, MYC, MYCL, MYCN, MYD88, NBN, NF1/2, NFE2L2, NOTCH1-3, NRAS, NTRK1-3, PALB2, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PIK3R1, PMS2, POLE, PPARG, PPP2R1A, PTCH1, PTEN, PTPN11, RAC1, RAF1, RAD50/51/51B/51C/51D, RB1, RET, RHEB, RHOA, RICTOR, RNF43, ROS1, SETD2, SLX4, SMAD4, SMARCB1, SMO, SPOP, SRC, STAT3, STK11, TERT, TOR1, TP53, TSC1-2, U2AF1, XPO1

AKT2, ALK, AR, AXL, BRAF, BRCA1, BRCA2, CDKN2A, EGFR, ERBB2, ERBB4, ERG, ESR1, ETV1, ETV4, ETV5, FGFR1-3, FGR, FLT3, JAK2, KRAS, MDM4, MET, MYB, MYBL1, NF1, NOTCH1, NOTCH4, NRG1, NTRK1-3, NUTM1, PDGFRA, PDGFRB, PIK3CA, PPARG, PRKACA, PRKACB, PTEN, RAD51B, RAF1, RB1, REL, RET, ROS1, RSPO2, RSPO3 und TERT

324 Gene

ABL1, ACVR1B, AKT1-3, ALK, ALOX12B, AMER1, APC, AR, ARAF, ARFRP1, ARID1A, ASXL1, ATM, ATR, ATRX, AURKA, AURKB, AXIN1, AXL, BAP1, BARD1, BCL2, BCL2L1-2, BCL6, BCOR, BCORL1, BRAF, BRCA1-2, BRC4, BRIP1, BTG1, BTK, C11orf30, CALR, CARD11, CASP8, CBF, CBL, CCND1-3, CCNE1, CD22, CD274, CD70, CD79A, CD79B, CDC73, CDH1, CDK12/4/6/8, CDKN1A/B, CDKN2A/B/C, CEBPA, CHEK1-2, CIC, CREBBP, CRKL, CSF1R, CSF3R, CTCF, CTNNA1, CTNNB1, CUL3, CXCR4, CYP17A1, DAXX, DDR1/2, DIS3, DNMT3A, DOT1L, EED, EGFR, EP300, EPHA3, EPHB1/4, ERBB2-4, ERCC4, ERG, ERF1, ESR, EZH2, FAM46C, FANCA, FANCC, FANCG, FANCL, FAS, FBXW7, FGF10/12/14/19/23/3/4/6, FGFR1-4, FH, FLCN, FLT1/3, FOXL2, FUBP1, GABRA6, GATA3/4/6, GID4, GNA11/13, GNAQ, GNAS, GRM3, GSK3B, H3F3A, HDAC1, HGF, HNF1A, HRAS, HSD3B1, ID3, IDH1-2, IGF1R, IKBKE, IKZF1, INPP4B, IRF2/4, IRS2, JAK1-3, JUN, KDM5A/C, KDM6A, KDR, KEAP1, KEL, KIT, KMT2A/D, KRAS, LTK, LYN, MAF, MAP2K1-4, MAP3K1/13, MAPK1, MCL1, MDM2/4, MED12, MEF2B, MEN1, MERTK, MET, MITF, MKNK1, MLH1, MPL, MRE11A, MSH2/3/6, MST1R, MTAP, MTOR, MUTYH, MYC, MYCL, MYCN, MYD88, NBN, NF1/2, NFE2L2, NFKBIA, NKX2-1, NOTCH1-3, NPM1, NRAS, NT5C2, NTRK1-3, P2RY8, PALB2, PARK2, PARP1-3, PAX5, PBRM1, PDCD1, PDCD1LG2, PDGFRA/B, PDK1, PIK3C2B/G, PIK3CA/B, PIK3R1, PIM1, PMS2, POLD1, POLE, PPARG, PPP2R1A, PPP2R2A, PRDM1, PRKAR1A, PRKCI, PTCH1, PTEN, PTPN11, PTPRO, QKI, RAC1, RAD21/51/51B/51C/51D//52/54L, RAF1, RARA, RB1, RBM10, REL, RET, RICTOR, RNF43, ROS1, RPTOR, SDHA/B/C/D, SETD2, SF3B1, SGK1, SMAD2/4, SMARCA4, SMARCB1, SMO, SNCAIP, SOCS1, SOX2/9, SPEN, SPOP, SRC, STAG2, STAT3, STK11, SUFU, SYK, TBX3, TEK, TET2, TGFB2, TIPARP, TNFAIP3, TNFRSF14, TP53, TSC1-2, TYRO3, U2AF1, VEGFA, VHL, WHSC1, WHSC1L1, WT1, XPO1, XRCC2, ZNF217/703

ALK, BCL2, BCR, BRAF, BRCA1-2, CD74, EGFR, ETV4/5, FGFR1-3, KIT, KMT2A, MSH2, MYB, MYC, NOTCH2, NTRK1-2, NUTM1, PDGFRA, RAF1, RARA, RET, ROS1, RSPO2, SDC4, SLC34A2, TERC, TERT, TMPRSS2