

ANA LISI

Patologia molecolare



VIOLLIER

Cancer Panels

Oncomine™ Comprehensive Assay v3 DNA, Thermo Fisher – 146 geni

Mutazioni hotspot

AKT1, AKT2, AKT3, ALK, AR, ARAF, AXL, BRAF, BTK, CBL, CCND1, CDK4, CDK6, CHEK2, CSF1R, CTNNB1, DDR2, EGFR, ERBB2 (=HER2), ERBB3, ERBB4, ERCC2, ESR1, EZH2, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, FOXL2, GATA2, GNA11, GNAQ, GNAS, H3F3A, HIST1H3B, HNF1A, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KDR, KIT, KNSTRN, KRAS, MAGOH, MAP2K1 (=MEK1), MAP2K2 (=MEK2), MAP2K4, MAPK1, MAX, MDM4, MED12, MET, MTOR, MYC, MYCN, MYD88, NFE2L2, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PPP2R1A, PTPN11, RAC1, RAF1, RET, RHEB, RHOA, ROS1, SF3B1, SMAD4, SMO, SPOP, SRC, STAT3, TERT, TOP1, U2AF1, XPO1

Sequenziamento completo del gene

ARID1A, ATM, ATR, ATRX, BAP1, BRCA1, BRCA2, CDK12, CDKN1B, CDKN2A, CDKN2B, CHEK1, CREBBP, FANCA, FANCD2, FANCI, FBXW7, MLH1, MRE11A, MSH2, MSH6, NBN, NF1, NF2, NOTCH1, NOTCH2, NOTCH3, PALB2, PIK3R1, PMS2, POLE, PTCH1, PTEN, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RB1, RNF43, SETD2, SLX4, SMARCA4, SMARCB1, STK11, TP53, TSC1, TSC2

Amplificazione del gene

AKT1, AKT2, AKT3, ALK, AR, AXL, BRAF, CCND1, CCND2, CCND3, CCNDE1, CDK2, CDK4, CDK6, CDKN2A, CDKN2B, EGFR, ERBB2, ESR1, FGF19, FGF3, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, IGF1R, KIT, KRAS, MDM2, MDM4, MET, MYC, MYCL, MYCN, NTRK1, NTRK2, NTRK3, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PPARG, RICTOR, TERT, TSC1, TSC2

Oncomine™ Comprehensive Assay RNA, Thermo Fisher – 52 partner di fusione

Geni di fusione

AKT2, ALK, AR, AXL, BRAF, BRCA1, BRCA2, CDKN2A, EGFR, ERBB2, ERBB4, ERG, ESR1, ETV1, ETV4, ETV5, FGFR1, FGFR2, FGFR3, FGR, FLT3, JAK2, KRAS, MDM4, MET, MYB, MYBL1, NF1, NOTCH1, NOTCH4, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, PDGFRA, PDGFRB, PIK3CA, PPARG, PRKACA, PRKACB, PTEN, RAD51B, RAF1, RB1, RELA, RET, ROS1, RSPO2, RSPO3, TERT

Oncomine™ Focus Assay DNA, Thermo Fisher – 50 geni

Mutazioni hotspot

AKT1, ALK, AR, BRAF, CDK4, CTNNB1, DDR2, EGFR, ERBB2 (=HER2), ERBB3, ERBB4, ESR1, FGFR2, FGFR3, GNA11, GNAQ, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KIT, KRAS, MAP2K1 (=MEK1), MAP2K2 (= MEK2), MET, MTOR, NRAS, PDGFRA, PIK3CA, RAF1, RET, ROS1, SMO1

Amplificazione del gene

ALK, AR, BRAF, CCND1, CDK4, CDK6, EGFR, ERBB2, FGFR1, FGFR2, FGFR3, FGFR4, KIT, KRAS, MET, MYC, MYCN, PDGFRA, PIK3CA

Oncomine™ Focus Assay RNA, Thermo Fisher – 23 partner di fusione

Geni di fusione

ABL1, ALK, AKT3, AXL, BRAF, EGFR, ERBB2, ERG, ETV1, ETV4, ETV5, FGFR1, FGFR2, FGFR3, MET, NTRK1, NTRK2, NTRK3, PDGFRA, PPARG, RAF1, RET, ROS1

Oncomine™ Tumor Mutation Load Assay (TMB), Thermo Fisher

NGS colon

Mutazioni hotspot

KRAS, BRAF, NRAS, PIK3CA

NGS polmone

Mutazioni hotspot

EGFR, KRAS, BRAF, HER2, MET

Geni di fusione

ALK1, ROS1, RET, NTRK1, MET Exon 14 skipping

NGS melanoma

Mutazioni hotspot

BRAF, KIT, NRAS, HRAS

NGS GIST

Mutazioni hotspot

KIT, PDGFRA, BRAF

NGS tiroide

Mutazioni hotspot

AKT1, BRAF, CTNNB1, HRAS, KRAS, NRAS, PIK3CA, PTEN, RET, TERT, TP53

Geni di fusione

NTRK1, NTRK2, NTRK3, PPARG, RET

NGS prostata

Sequenziamento completo del gene

BRCA1, BRCA2, ATM, ATR, CDK12, CHEK1, CHEK2, FANCA, NBN, PALB2, RAD50, RAD51, RAD51B, RAD51C, RAD51D

NGS endometrio

Sequenziamento completo del gene

POLE, TP53

Ulteriori analisi:

BRCA1, BRCA2 Sequenziamento completo del gene

HRD-Test MyChoice Illumina TruSight Oncology 500 HRD (validazione in corso)

FGFR2, FGFR3 Mutazioni hotspot

Instabilità dei microsatelliti (MSI) Analisi della lunghezza di frammento

Metilazione del promotore di MLH1 Analisi della lunghezza di frammento

PIK3CA, ESR1 Mutazioni hotspot

Biopsia liquida

Lista dei geni e provetta Streck per il prelievo su richiesta

Presso Viollier tutte le analisi elencate sono accreditate secondo la norma

SN EN ISO 15189.

Informazioni

Dr. rer. nat. Henriette Kurth, Specialista FAMH in medicina di laboratorio, Responsabile Patologia molecolare
Dr. med. Katharina Marston, FMH patologia, patologia molecolare, Responsabilità medica Patologia molecolare

PD Dr. med. Andreas Zettl, FMH patologia, Responsabile Patologia, Candidato patologia molecolare
Dr. med. Sophie Diebold Berger, FMH patologia, citopatologia, Responsabile Viollier Weintraub SA