ANA LISI

Patologia molecolare

VIOLLIER

Cancer Panels

Oncomine™ Comprehensive Assay v3 DNA, Thermo Fisher – 145 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, HNF1A, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KDR, KIT, KNSTRN, KRAS, MAGOH, MAP2K1 (=MEK1), MAP2K2 (=MEK2), MAP2K4, MAPK1, MAX, MDM2, MDM4, MED12, MET, MTOR, MYC, MYCL, 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, MRE11, 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

Alterazioni del numero di copie (CNVs)

AKT1, AKT2, AKT3, ALK, AR, AXL, BRAF, CCND1, CCND2, CCND3, CCNE1, CDK2, CDK4, CDK6, 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

Oncomine™ Comprehensive Assay RNA, Thermo Fisher – 51 partner di fusione

Fusioni di geni

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™ Precision Assay DNA, Thermo Fisher – 46 geni

Mutazioni hotspot

AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CDKN2A, CHEK2, CTNNB1, EGFR, ERBB2 (=HER2), ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1 (=MEK1), MAP2K2 (= MEK2), MET, MTOR, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, SMO, TP53

Alterazioni del numero di copie (CNVs)

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

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

Fusioni di geni

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

NGS Carcinoma colorettale

Mutazioni hotspot

KRAS, NRAS, BRAF, PIK3CA

NGS Carcinoma polmonare

Mutazioni hotspot

EGFR, KRAS, BRAF, HER2, MET

Fusioni di geni

ALK1, ROS1, RET, NTRK1, NTRK2, NTRK3, MET Exon-14-Skipping

NGS melanoma

Mutazioni hotspot

BRAF, KIT, NRAS, HRAS

NGS tumore stromale gastrointestinale (GIST)

Mutazioni hotspot

KIT, PDGFRA

NGS Carcinoma della tiroide

Mutazioni hotspot

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

Fusioni di geni

RET, PPARG, NTRK1, NTRK2, NTRK3

NGS Carcinoma dell'endometrio

Sequenziamento completo del gene

POLE, TP53, MLH1, MSH2, MSH6, PMS2

NGS Carcinoma uroteliale

Mutazioni hotspot e fusioni

FGFR1, FGFR2, FGFR3, FGFR4

Sequenziamento completo del gene

TP53

NGS Carcinoma ovarico
TruSight Oncology 500 HRD powered by Myriad, Illumina
Mutazioni in geni associati alla BRCAness incl. BRCA1, BRCA2, PALB2
Valutazione del Genomic Instability Score (GIS)

NGS Carcinoma mammario pannello grande
TruSight Oncology 500, Illumina
Mutazioni in geni associati alla BRCAness incl. BRCA1, BRCA2, PALB2
Incl. PIK3CA, AKT1, PTEN, ESR1

NGS Carcinoma mammario pannello piccolo
Oncomine™ Comprehensive Assay v3 DNA, Thermo Fisher
PIK3CA, AKT1, PTEN, ESR1

Biopsia liquida Carcinoma mammario
Oncomine™ Precision Assay GX, Thermo Fisher
ESR1, PIK3CA, AKT1

NGS Carcinoma prostatico
TruSight Oncology 500, Illumina
Mutazioni in geni associati alla BRCAness incl. BRCA1, BRCA2, PALB2
Incl. analisi dell'instabilità microsatellitare e CDK12

Analisi supplementari:
BRCA1, BRCA2 Sequenziamento completo del gene
Genomic Instability Score (GIS) TruSight Oncology 500 HRD powered by Myriad
Instabilità dei microsatelliti (MSI) Analisi della lunghezza di segmento
Metilazione del promotore di MLH1 Analisi della lunghezza di segmento
Carico mutazionale del tumore (TMB) TruSight Oncology 500, Illumina

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 Biologia molecolare
Dr. med. Katharina Marston, FMH patologia, patologia molecolare, Resp. medicia patologia molecolare
Dr. med. Sophie Diebold Berger, FMH patologia, citopatologia, Responsabile Viollier Genève SA Patologia
PD Dr. med. Andreas Zettl, FMH patologia, candidato patologia molecolare, Responsabile Patologia

viollier.ch