

Analisi

Patologia molecolare

Cancer Panels

Oncomine™ Comprehensive Assay v3 DNA, Thermo Fisher – 145 gènes

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
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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
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NGS Carcinoma coloretale

Mutazioni hotspot	KRAS, NRAS, BRAF, PIK3CA
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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
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NGS tumore stromale gastrointestinale (GIST)

Mutazioni hotspot	KIT, PDGFRA
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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
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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)
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NGS Carcinoma mammario pannello grande

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

Oncomine™ Comprehensive Assay v3 DNA, Thermo Fisher	PIK3CA, AKT1, PTEN, ESR1
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Biopsia liquida Carcinoma mammario

Oncomine™ Precision Assay GX, Thermo Fisher	ESR1, PIK3CA, AKT1
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NGS Carcinoma prostatico

TruSight Oncology 500, Illumina	Mutazioni in geni associati alla BRCAness incl. BRCA1, BRCA2, ATM Incl. analisi dell'instabilità microsatellitare e CDK12
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Analisi supplementari

BRCA1, BRCA2 Sequenziamento completo del gene

Genomic Instability Score (GIS) TruSight Oncology 500 HRD powered by Myriad

Mikrosatelliten-Instabilität (MSI) Analisi della lunghezza di segmento

MLH1-Promotormethylierung 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

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