Istituto Cantonale di Patologia EOC
Servizio di Patologia e Genetica Molecolare

Lista pannelli NGS e geni inclusi

Oncomine™ Comprehensive Assay v3 **146 geni**	Alterazioni identificate*: SNV, INDEL
ARAF, ARID1A, ATM, ATR, ATRX, BAP1, BRCA1, BRAC2, BTK, CBL, CDK12, CDKN1B, CDKN2A, CDKN2B,CHEK1, CHEK2, CREBBP, CSF1R, CTNNB1, DDR2, ERBB3, ERBB4, ERCC2, EZH2, FANCA, FANCD2, FANCI, FBXW7, FOXL2, GATA2, GNA11, GNAQ, GNAS, H3F3A, HIST1H3B, HNF1A, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KDR, KNSTRN, MAGOH, MAP2K1, MAP2K2, MAP2K4, MAPK1, MAX, MED12, MLH1, MRE11A, MSH2, MSH6, MTOR, MYD88, NBN, NF1, NF2, NFE2L2, NOTCH1, NOTCH2, NOTCH3, NRAS, PALB2, PIK3R1, PMS2, POLE, PP2R1A, PTCH1, PTEN, PTPN1, RAC1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAF1, RB1, RET, RHEB, RHOA, RNF43,ROS1, SETD2, SF3B1, SLX4, SMAD4, SMARCA4, SMARCB1, SMO, SPOP, SRC, STAT3, STK11, TOP1, TP53, TSC1, TSC2, U2AF1, XPO	
	SNV, INDEL, CNV
AKT1, AKT2, AKT3, ALK, AR, AXL, BRAF, CCND1, CDK4, CDK6, EGFR, ERBB2, ESR1, FGFR1 FGFR2, FGFR3, FGFR4, FLT3, KIT, KRAS, MDM4, MET, MYC, MYCN, NTRK1, NTRK2, NTRK3, PDGFRA, PDGFRB, PIK3CA, PIK3CB, TERT	
	CNV
CCND2, CCND3, CCNE, CDK2, FGF19, FGF3 IGF1R, MDM2, MYCL, PPARG, RICTOR	

Ion AmpliSeq™ Cancer Hotspot Panel v2 **50 geni**	Alterazioni identificate*: SNV, INDEL
ABL1, AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CSFR1, CTNNB1, EGFR, ERBB2, ERBB4, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, FLT3, GNA11, GNAS, GNAQ, HNF1A, HRAS, IDH1, IDH2, JAK2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53, VHL	

Ion AmpliSeq™ Lung Cancer Panel v2 **22 geni**	Alterazioni identificate*: SNV, INDEL
AKT1, ALK, BRAF, CTNNB1, DDR2, EGFR, ERBB2, ERBB4, FBXW7, FGFR1, FGFR2, FGFR3, KRAS, MAP2K1, MET, NRAS, NOTCH, PIK3CA, PTEN, STK11, SMAD4, TP53	

Endometrial Custom Panel ***6 geni***	Alterazioni identificate*: SNV, INDEL
POLE, KRAS, PTEN, PIK3CA, TP53, CTNNB1	

Oncomine™ BRCA Research Assay **2 geni**	Alterazioni identificate*: SNV, INDEL
BRCA1, BRCA2	

* SNV: single nucleotide variant (mutazioni a singolo nucleotide), INDEL: inserzioni e delezioni di corte regioni nucleotidiche, CNV: copy number variation (amplificazioni e copy number gain)

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Lista pannelli NGS e geni inclusi

Oncomine™ Tumor Mutation Load Assay 409 geni	Alterazioni identificate*: SNV, INDEL, TML

ABL1, ABL2, ACVR2A, ADAMTS20, AFF1, AFF3, AKAP9, AKT1, AKT2, AKT3, ALK, APC, AR, ARID1A, ARID2, ARNT, ASXL1, ATF1, ATM, ATR, ATRX, AURKA, AURKB, AURKC, AXL, BAI3, BAP1, BCL10, BCL11A, BCL11B, BCL2, BCL2L1, BCL2L2, BCL3, BCL6, BCL9, BCR, BIRC2, BIRC3, BIRC5, BLM, BLNK, BMPR1A, BRAF, BRD3, BTK, BUB1B, CARD11, CASC5, CBL, CCND1, CCND2, CCNE1, CD79A, CD79B, CDC73, CDH1, CDH11, CDH2, CDH20, CDH5, CDK4, CDK6, CDK8, CDK12, CDKN2A, CDKN2B, CDKN2C, CEBPA, CHEK1, CHEK2, CIC, CKS1B, CMPK1, COL1A1, CRBN, CREBBP, CREB1, CRKL, CRTC1, CSF1R, CSMD3, CTNNA1, CTNNB1, CYLD, CYP2C19, CYP2D6, DAXX, DCC, DDB2, DDIT3, DDR2, DEK, DICER1, DNMT3A, DPYD, DST, EML4, EGFR, ERBB2, ERBB3, ERBB4, EP300, EP400, EPHA3, EPHA7, EPHB1, EPHB4, EPHB6, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ESR1, ETS1, ETV1, ETV4, EXT1, EXT2, EZH2, FAM123B, FANCA, FANCC, FANCD2, FANCF, FANCG, FANCJ, FAS, FBXW7, FGFR1, FGFR2, FGFR3, FGFR4, FH, FLCN, FLI1, FLT1, FLT34, FLT4, FN1, FOXL2, FOXO1, FOXO3, FOXP1, FOXP4, FZR1, G6PD, GATA1, GATA2, GATA3, GDNF, GNA11, GNAQ, GNAS, GPR124, GRM8, GUCY1A2, HCAR1, HIF1A, HLF, HNF1A, HOOK3, HRAS, HSP90AA1, HSP90AB1, ICK, IDH1, IDH2, IGF1R, IGF2, IGF2R, IKBKB, IKBKE, IKZF1, IL2, IL21R, IL6ST, IL7R, ING4, IRF4, IRS2, ITGA10, ITGA9, ITGB2, ITGB3, JAK1, JAK2, JAK3, JUN, KAT6A, KAT6B, KDM5C, KDM6A, KDR, KEAP1, KIT, KLF6, KRAS, LAMP1, LCK, LIFR, LPHN3, LPP, LRP1B, LTF, LTK, MAF, MAFB, MAGEA1, MAGI1, MALT1, MAML2, MAP2K1, MAP2K2, MAP2K4, MAP3K7 MAPK1, MAPK8, MARK1, MARK4, MBD1, MCL1, MDM2, MDM4, MEN1, MET, MITF, MLH1, MLL, MLL2, MLLT10, MMP2, MN1, MPL, MRE11A, MSH2, MSH6, MTOR, MTR, MTRR, MUC1, MUTYH, MYB, MYC, MYCN, MYCL1, MYD88, MYH11, MYH9, NBN, NCOA1, NCOA2, NCOA4, NF1, NF2, NFE2L2, NFKB1, NFKB2, NIN, NKX2-1, NLRP1, NOTCH1, NOTCH2, NOTCH4, NPM1, NRAS, NSD1, NTRK1, NTRK3, NUMA1, NUP214, NUP98, PALB2, PAK3, PARP1, PAX3, PAX5, PAX7, PAX8, PBRM1, PBX1, PDE4DIP, PDGFB, PDGFRA, PDGFRB, PIK3CA, PIK3CB, PIK3R1, PER1, PGAP3, PHOX2B, PIK3C2B, PIK3CD, PIK3CG, PIK3R2, PIM1, PKHD1, PLAG1, PLCG1, PLEKHG5, PML, PMS1, PMS2, POT1, POU5F1, PPARG, PPP2R1A, PRDM1, PRKAR1A, PRKDC, PSIP1, PTCH1, PTEN, PTGS2, PTPN11, PTPRD, PTPRT, RAD50, RAF1, RALGDS, RARA, RB1, RECQL4, REL, RET, RHOH, RNASEL, RNF2, RNF213, ROS1, RPS6KA2, RRM1, RUNX1, RUNX1T1, SAMD9, SBDS, SDHA, SDHB, SDHC, SDHD, SEPT9, SETD2, SF3B1, SGK1, SH2D1A, SMAD2, SMAD4, SMARCA4, SMARCB1, SMO, SMUG1, SOCS1, SOX11, SOX2, SRC, SSX1, STK11, STK36, SUFU, SYK, SYNE1, TAF1, TAF1L, TAL1, TBX22, TCF12, TCF3, TCF7L1, TCF7L2, TCL1A, TET1, TET2, TFE3, TGFBR2, TGM7, THBS1, TIMP3, TLR4, TLX1, TNFAIP3, TNFRSF14, TNK2, TOP1, TP53, TPR, TRIM24, TRIM33, TRIPO11, TRRAP, TSC1, TSHR, UBR5, UGT1A1, USP9X, VHL, WAS, WHSC1, WRN, XPA, XPC, XPO1, XRCC2, ZNF384, ZNF521

Archer FusionPlex Custom panel 65 geni	Alterazioni identificate: geni di fusione

ACVR2A, ALK, BCOR, BRAF, CAMTA1, CCNB3, CHMP2A, CIC, CSF1, EGFR, EPC1, ERG, ETV1, EWSR1, FGF1, FGFR1, FGFR2, FGFR3, FGR, FOXO1, FUS, GLI1, GPI, GRM1, HMGA2, IGF1R, JAZF1, MAML2, MEAF6, MET, MGEA5, MKL2, MSANTD3, MYB, MYBL1, NCOA2, NFATC2, NOTCH1, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, PAX5, PDGFB, PHF1, PIK3CA, PLAG1, PPARG, PRKD1, RAB7A, RAF1, RET, ROS1, SRF, SS18, STAT6, TAF15, TCF12, TFE3, TFG, TMPRSS2, USP6, VCP, YWHAE

* SNV: single nucleotide variant (mutazioni a singolo nucleotide), INDEL: inserzioni e delezioni di corte regioni nucleotidiche, TML: Tumor Mutation Load

Lista pannelli NGS e geni inclusi

Archer FusionPlex Lung **14 geni**	Alterazioni identificate: geni di fusione
ALK, BRAF, EGFR, FGFR1, FGFR2, FGFR3, KRAS, MET, NRG1, NTRK1, NTRK2, NTRK3, RET, ROS1	

Oncomine™ Colon cfDNA Assay **14 geni**	Alterazioni identificate*: SNV, INDEL
AKT1, APC, BRAF, CTNNB1, EGFR, ERBB2, FBXW7, GNAS, KRAS, MAP2K1 (MEK1), NRAS, PIK3CA, SMAD4, TP53	

Oncomine™ Lung cfDNA Assay **11 geni**	Alterazioni identificate*: SNV, INDEL
ALK, BRAF, EGFR, ERBB2, KRAS, MAP2K1(MEK1), MET, NRAS, PIK3CA, ROS1, TP53	

Lymphoma Custom Panel **172 geni**	Alterazioni identificate*: SNV, INDEL
ACTB, ARID1A, ARID5B, ATM, B2M, BCL10, BCL11A, BCL2, BCL6, BCL7A, BCOR, BIRC3, BRAF, BTG1, BTG2, BTK, CALR , CARD11, CCND1, CCND3, CCR4, CD19, CD274, CD28, CD58, CD70, CD79A, CD79B, CD83, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CIITA, CREBBP, CTNNB1, CTSS, CXCR4, CXCR5, DDX3X, DNMT3A, DTX1, EBF1, EGR2, EP300, EPHA7, ETS1, ETV6, EZH2, FAS, FBXW7, FLT3, FOXC1, FOXO1, FYN, GATA3, GNA13, GNAI2, GRHPR, HIST1H1B, HIST1H1C, HIST1H1D, HIST1H1E, HIST1H2AC, HIST1H2AM, HIST1H2BC, HIST1H2BK, HNF1B, HRAS, HVCN1, ID3, IDH1, IDH2, IGLL5, IKBKB, IKZF1, IKZF3, IL16, IL4R, IRF2BP2, IRF4, IRF8, ITGAE, ITPKB, JAK1, JAK2, JAK3, KIT, KLF2, KLHL6, KLHL14, KLHL21, KMT2C, KMT2D, KRAS, LTB, MAP2K1, MCL1, MEF2B, MEF2C, MGA, MS4A1, MYC, MYD88, NFKBIA, NFKBIE, NFKBIZ, NOL9, NOTCH1, NOTCH2, NRAS, P2RY8, PAX5, PCBP1, PHF6, PIK3CA, PIK3CD, PIK3R1, PIM1, PLCG1, PLCG2, POT1, PPM1D, PPP1R9B, PRDM1, PRKCB, PTEN, PTPN1, PTPN11, PTPRD, PTPRK, RB1, RELN, RERE, RHOA, RPS15, RRAGC, S1PR2, SAMHD1, SETD1B, SETD2, SF3B1, SGK1, SMARCA4, SOCS1, SOCS3, SPEN, STAT3, STAT5B, STAT6, TBL1XR1, TCF3, TET2, TMEM30A, TMSB4X, TNFAIP3, TNFRSF14, TOX, TP53, TRAF2, TRAF3, TRIP12, TRRAP, UBE2A, VMP1, VPS13B, WEE1, WHSC1, XPO1, ZC3H12A, ZEB2, ZFP36L1.	

Per ulteriori informazioni:
https://www.thermofisher.com/ch/en/home.html
https://archerdx.com/

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