**Istituto Cantonale di Patologia**
Servizio di patologia molecolare



# LISTA PANNELLI NGS E GENI INCLUSI

| **Oncomine™ Comprehensive Assay v3**<br>**146 geni** | Alterazioni identificate*:<br>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**<br>**50 geni** | Alterazioni identificate*:<br>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™ Colon and Lung Cancer Research Panel**<br>**22 geni** | Alterazioni identificate*:<br>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***<br>***6 geni*** | Alterazioni identificate*:<br>SNV, INDEL |
|---|---|
| *POLE, KRAS, PTEN, PIK3CA, TP53, CTNNB1* | |

| **Oncomine™ BRCA Research Assay**<br>**2 geni** | Alterazioni identificate*:<br>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**<br>**409 geni** | Alterazioni identificate*:<br>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**<br>**65 geni** | Alterazioni identificate:<br>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


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NEXT GENERATION SEQUENCING

# LISTA PANNELLI NGS E GENI INCLUSI

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

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

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

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

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


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