# JAX SomaticSeq Technical Specifications

The Jackson Laboratory

## INTENDED USE

JAX SomaticSeq is a qualitative next generation sequencing based assay that uses targeted selection of DNA from 517 genes of interest, and RNA from 55 known drivers, for a total of 1.94Mb panel size. JAX SomaticSeq detects genomic signatures including microsatellite instability (MSI) and tumor mutational burden (TMB) using DNA and RNA isolated from formalin-fixed paraffin embedded (FFPE) tissue specimens in a single, streamlined workflow. Over 1,000 clinical trials are available for the range of variants targeted by the assay. JAX SomaticSeq was developed with the Illumina TruSight Oncology 500 Assay and its performance characteristics determined by The Jackson Laboratory. It has not been cleared or approved by the US Food and Drug Administration.

## GENE LIST

| Small Variants Only | | Small Variants & CNVs | | Small Variants & Fusions | | Small Variants, CNVs, & Fusions | |
|---|---|---|---|---|---|---|---|
| ABL1 | CD274 | EPHB1 | GATA6 | KDM5A | NFE2L2 | PRKN (PARK2) | SOX10 |
| ABL2 | CD276 | ERBB2 (HER2) | GEN1 | KDM5C | NFKBIA | PRSS8 | SOX17 |
| ABRAXAS1 (FAM175A) | CD74 | ERBB3 | GID4 | KDM6A | NKX2-1 | PTCH1 | SOX2 |
| ACVR1 | CD79A | ERBB4 | GLI1 | KDR | NKX3-1 | PTEN | SOX9 |
| ACVR1B | CD79B | ERCC1 | GNA11 | KEAP1 | NOTCH1 | PTPN11 | SPEN |
| ADGRA2 (GPR124) | CDC73 | ERCC2 | GNA13 | KEL | NOTCH2 | PTPRD | SPOP |
| AKT1 | CDH1 | ERCC3 | GNAQ | KIF5B | NOTCH3 | PTPRS | SPTA1 |
| AKT2 | CDK12 | ERCC4 | GNAS | KIT | NOTCH4 | PTPRT | SRC |
| AKT3 | CDK4 | ERCC5 | GPS2 | KLF4 | NPM1 | QKI | SRSF2 |
| ALK | CDK6 | ERG | GREM1 | KLHL6 | NRAS | RAB35 | STAG1 |
| ALOX12B | CDK8 | ERRFI1 | GRIN2A | KMT2A (MLL) | NRG1 | RAC1 | STAG2 |
| AMER1 (FAM123B) | CDKN1A | ESR1 | GRM3 | KRAS | NSD1 | RAD21 | STAT3 |
| ANKRD11 | CDKN1B | ETS1 | GSK3B | LAMP1 | NTRK1 | RAD50 | STAT4 |
| ANKRD26 | CDKN2A | ETV1 | H1-2 (HIST1H1C) | LATS1 | NTRK2 | RAD51 | STAT5A |
| APC | CDKN2B | ETV4 | H2BC5 (HIST1H2BD) | LATS2 | NTRK3 | RAD51B | STAT5B |
| AR | CDKN2C | ETV5 | H3-3A (H3F3A) | LMO1 | NUP93 | RAD51C | STK11 |
| ARAF | CEBPA | ETV6 | H3-3B (H3F3B) | LRP1B | NUTM1 | RAD51D | STK40 |
| ARFRP1 | CENPA | EWSR1 | H3-4 (HIST3H3) | LYN | PAK1 | RAD52 | SUFU |
| ARID1A | CHD2 | EZH2 | H3-5 (H3F3C) | LZTR1 | PAK3 | RAD54L | SUZ12 |
| ARID1B | CHD4 | FANCA | H3C1 (HIST1H3A) | MAGI2 | PAK5 (PAK7) | RAF1 | SYK |
| ARID2 | CHEK1 | FANCC | H3C10 (HIST1H3H) | MALT1 | PALB2 | RANBP2 | TAF1 |
| ARID5B | CHEK2 | FANCD2 | H3C11 (HIST1H3I) | MAP2K1 | PARP1 | RARA | TBX3 |
| ASXL1 | CIC | FANCE | H3C12 (HIST1H3J) | MAP2K2 | PAX3 | RASA1 | TCF3 |
| ASXL2 | COP1 (RFWD2) | FANCF | H3C13 (HIST2H3D) | MAP2K4 | PAX5 | RB1 | TCF7L2 |
| ATM | CREBBP | FANCG | H3C14 (HIST2H3C) | MAP3K1 | PAX7 | RBM10 | TENT5C (FAM46C) |
| ATR | CRKL | FANCI | H3C15 (HIST2H3A) | MAP3K13 | PAX8 | RECQL4 | TERC |
| ATRX | CRLF2 | FANCL | H3C2 (HIST1H3B) | MAP3K14 | PBRM1 | REL | TERT |
| AURKA | CSF1R | FAS | H3C3 (HIST1H3C) | MAP3K4 | PDCD1 | RET | TET1 |
| AURKB | CSF3R | FAT1 | H3C4 (HIST1H3D) | MAPK1 | PDCD1LG2 | RHEB | TET2 |
| AXIN1 | CSNK1A1 | FBXW7 | H3C6 (HIST1H3E) | MAPK3 | PDGFRA | RHOA | TFE3 |
| AXIN2 | CTCF | FGF1 | H3C7 (HIST1H3F) | MAX | PDGFRB | RICTOR | TFRC |
| AXL | CTLA4 | FGF10 | H3C8 (HIST1H3G) | MCL1 | PDK1 | RIT1 | TGFBR1 |
| B2M | CTNNA1 | FGF14 | HGF | MDC1 | PDPK1 | RNF43 | TGFBR2 |
| BAP1 | CTNNB1 | FGF19 | HNF1A | MDM2 | PGR | ROS1 | TMEM127 |
| BARD1 | CUL3 | FGF2 | HNRNPK | MDM4 | PHF6 | RPS6KA4 | TMPRSS2 |
| BBC3 | CUX1 | FGF23 | HOXB13 | MED12 | PHOX2B | RPS6KB1 | TNFAIP3 |
| BCL10 | CXCR4 | FGF3 | HRAS | MEF2B | PIK3C2B | RPS6KB2 | TNFRSF14 |
| BCL2 | CYLD | FGF4 | HSD3B1 | MEN1 | PIK3C2G | RPTOR | TOP1 |
| BCL2L1 | DAXX | FGF5 | HSP90AA1 | *MET | PIK3C3 | RUNX1 | TOP2A |
| BCL2L11 | DCUN1D1 | FGF6 | ICOSLG | MGA | PIK3CA | RUNX1T1 | TP53 |
| BCL2L2 | DDR2 | FGF7 | ID3 | MITF | PIK3CB | RYBP | TP63 |
| BCL6 | DDX41 | FGF8 | IDH1 | MLH1 | PIK3CD | SDHA | TRAF2 |
| BCOR | DHX15 | FGF9 | IDH2 | MLLT3 | PIK3CG | SDHAF2 | TRAF7 |
| BCORL1 | DICER1 | FGFR1 | IFNGR1 | MPL | PIK3R1 | SDHB | TSC1 |
| BCR | DIS3 | FGFR2 | IGF1 | MRE11 (MRE11A) | PIK3R2 | SDHC | TSC2 |
| BIRC3 | DNAJB1 | FGFR3 | IGF1R | MSH2 | PIK3R3 | SDHD | TSHR |
| BLM | DNMT1 | FGFR4 | IGF2 | MSH3 | PIM1 | SETBP1 | U2AF1 |
| BMPR1A | DNMT3A | FH | IKBKE | MSH6 | PLCG2 | SETD2 | VEGFA |
| BRAF | DNMT3B | FLCN | IKZF1 | MST1 | PLK2 | SF3B1 | VHL |
| BRCA1 | DOT1L | FLI1 | IL10 | MST1R | PMAIP1 | SH2B3 | VTCN1 |
| BRCA2 | E2F3 | FLT1 | IL7R | MTOR | PMS1 | SH2D1A | WT1 |
| BRD4 | EED | FLT3 | INHA | MUTYH | PMS2 | SHQ1 | XIAP |
| BRIP1 | EGFL7 | FLT4 | INHBA | MYB | PNRC1 | SLIT2 | XPO1 |
| BTG1 | *EGFR | FOXA1 | INPP4A | MYC | POLD1 | SLX4 | XRCC2 |
| BTK | EIF1AX | FOXL2 | INPP4B | MYCL (MYCL1) | POLE | SMAD2 | YAP1 |
| CALR | EIF4A2 | FOXO1 | INSR | MYCN | PPARG | SMAD3 | YES1 |
| CARD11 | EIF4E | FOXP1 | IRF2 | MYD88 | PPM1D | SMAD4 | ZBTB2 |
| CASP8 | ELOC (TCEB1) | FRS2 | IRF4 | MYOD1 | PPP2R1A | SMARCA4 | ZBTB7A |
| CBFB | EML4 | FUBP1 | IRS1 | NAB2 | PPP2R2A | SMARCB1 | ZFHX3 |
| CBL | EMSY (C11orf30) | FYN | IRS2 | NBN | PPP6C | SMARCD1 | ZNF217 |
| CCN6 (WISP3) | EP300 | GABRA6 | JAK1 | NCOA3 | PRDM1 | SMC1A | ZNF703 |
| CCND1 | EPCAM | GATA1 | JAK2 | NCOR1 | PREX2 | SMC3 | ZRSR2 |
| CCND2 | EPHA3 | GATA2 | JAK3 | NEGR1 | PRKAR1A | SMO | *EGFR exons 2-7 (vIII) & MET exon 14 skipping events also reported |
| CCND3 | EPHA5 | GATA3 | JUN | NF1 | PRKCI | SNCAIP | |
| CCNE1 | EPHA7 | GATA4 | KAT6A | NF2 | PRKDC | SOCS1 | |

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