Every patient is unique. So is every patient’s tumor. Advances in high-throughput molecular diagnostics now allow detection of the genetic alterations that are specific to an individual patient’s tumor. By identifying the oncogenic drivers, therapeutic options can be matched to the tumor’s biology.
TEST OVERVIEW

Gene fusions occur due to rearrangement of two separate genes resulting in a hybrid gene, often leading to the disabling of an intended gene product (protein) that is needed by the body, or the creation, and often overexpression, of an abnormal, typically harmful, chimeric protein. It can also lead to the fusing of an oncogene (tumor promoting gene) to a strong promoter, magnifying the impact of the cancer-causing gene. Fusion genes were originally associated with hematologic cancers but more than 300 gene fusions have been identified in almost every kind of cancer including solid tumors (sarcomas, carcinomas and tumors of the central nervous system). Identifying and characterizing these fusion genes therefore can have both diagnostic and therapeutic applications. The advent of next generation sequencing has enabled high-throughput, accurate detection of fusion genes.

The JAX FusionSeq™ assay is designed to detect fusions involving one or more of 53 genes known to be associated with various carcinomas, sarcomas and hematologic malignancies.

The FusionSeq™ test can be done in conjunction with the Cancer Treatment Profile™ (CTP) test, a broad-based NGS analysis of 258 clinically relevant genes (for SNPs, CNVs and micro InDels), the ActionSeq™ test (for SNPs CNVs and micro InDels), or as a stand-alone test.

METHODS

Total RNA is extracted from macro dissection-enriched FFPE tissue sections, followed by cDNA synthesis and amplification to generate a minimum of 1.5 million reads per sample. The fusion detection limit is 5% of wild-type with a minimum of ≥15 supporting reads and ≥5 unique start sites. Fusion analysis is performed using the Archer™ Analysis Software, which aligns against the hg19 human genome build.

SPECIMEN REQUIREMENTS

- Formalin-fixed, paraffin-embedded (FFPE) material only.
- One representative hematoxylin and eosin (H&E) stained slide and 5 to 10 adjacent unstained 5 um sections on uncoated, unbaked slides. We also accept tumor blocks.
- Any solid tumor, primary or metastatic tissue. The area of highest tumor cell content should be a minimum of 3 x 3 mm.

GENE LIST

AKT3, ALK, ARHGAP26, AXL, BRAF, BRD3, BRD4, EGFR, ERG, ESR1, ETV1, ETV4, ETV5, ETV6, EWSR1, FGFR1, FGFR2, FGFR3, FGR, INSR, MAML2, MAST1, MAST2, MET, MSMB, MUSK, MYB, NOTCH1, NOTCH2, NRG1, NTRK1, NTRK2, NTRK3, NUMBL, NUTM1, PDGFRA, PDGFRB, PIK3CA, PKN1, PPARG, PRKCA, PRKCB, RAF1, RELA, RET, ROS1, RSPO2, RSPO3, TERT, TFE3, TFEB, THADA, TMPRSS2

REFERENCES


jax.org/fusionseq