

## NGS Sarcoma Fusion Panel

Effective Date: December 20<sup>th</sup>, 2022

The Molecular Pathology Laboratory at Beaumont Health will offer next generation sequencing that enables genomic detection of fusions in sarcomas. This method permits the identification of the presence of a rearrangement even if the fusion partner is not known and is not included in the panel. This test is designed for sequencing RNA for the detection of gene fusions/rearrangements in the following genes (specific targeted regions of each gene can be found in the lab test directory):

*ALK, BCOR, BRAF, CAMTA1, CCNB3, CIC, CSF1, CTNNB1, EGFR, EPC1, ERG, ESR1, ETV1, ETV4, ETV5, ETV6, EWSR1, FGFR1, FGFR2, FGFR3, FOS, FOSB, FOXO1, FUS, GLI1, HMGA2, JAZF1, MBTD1, MDM2, MEAF6, MET, MGEA5, MKL2, MYOD1, NCOA1, NCOA2, NCOA3, NR4A3, NTRK1, NTRK2, NTRK3, NUTM1, PAX3, PDGFB, PDGFRA, PHF1, PLAG1, PRKCA, PRKCB, PRKCD, RAF1, RET, ROS1, SS18, STAT6, TAF15, TCF12, TFE3, TFG, USP6, VGLL2, YAP1, YWHAE.*

This test will be ordered by the pathologist to supplement morphological and immunohistochemical results with molecular results that may identify fusions necessary for precise diagnosis of sarcomas.

### Synonyms:

Sarcoma fusion, next generation sequencing, NGS, sarcoma panel.

### Specimen Collection Criteria:

Paraffin-embedded tissue block must be submitted with corresponding H&E slide. Unstained sections of 5- $\mu$ m thickness mounted on glass slides can also be used (minimum 5 sections for large tissue and 10 sections for small tissue such as core biopsy). Tissue should be well fixed and well processed. Recommended tissue size 5.0 mm<sup>2</sup>. Minimum tumor cellularity is 30%.

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- RNA will be assessed for quantity and quality. If either is deemed insufficient for analysis, testing will be cancelled with client notification.
- The specimen must be accompanied by a completed requisition and must contain the patient's name, date of birth, collection date, ordering physician, and source of specimen.

### Specimen Preparation for Courier Transport

Transport tissue blocks and unstained slides, at room temperature (20-26°C or 68-78.8°F).

### Rejection Criteria

- Tissue decalcified with agents other than Mol Decal (EDTA)
- Fixatives other than 10% neutral buffered formalin.
- Improper labeling or inadequate information.
- Less than 30% tumor cellularity
- Poor quality and/or quantity of extracted genomic RNA.
- Frozen specimens.
- Unlabeled samples.

Testing will be cancelled on specimens meeting the above criteria with client notification. Under certain circumstances (i.e., lack of alternative specimens), testing may proceed with approval from the medical director or designee.

### Performed

Once or twice per week, dependent upon test volume. Results available in 10-15 business days.

### Reference Range

No variants detected or likely benign variants detected.

### Test Methodology

Tissue section slides are reviewed by a pathologist and relevant tumor is selected for analysis. RNA is isolated from the sample and quantified. Recovered nucleic acid extracts are prepared for sequencing with the ArcherDx Invitae FusionPlex® Sarcoma v2 library preparation kit and sequenced on the Illumina® MiSeq™ instrument. Analysis is performed using Archer Analysis Unlimited (AAU) ® software.

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A personalized interpretive report is generated for each sample that lists the fusions detected in each gene. For each fusion reported, detailed information for the transcript and breakpoint are given. Fusions are classified based on a standardized classification scheme for somatic variants and provides interpretative comments for each variant of known significance.

### Interpretation

Test results should be interpreted in the context of clinical findings, tumor sampling, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory director.

### Test Code

SRCFS

### CPT Code

81455

G0452

If you have questions, please contact your Beaumont Laboratory Customer Service Department:

- Farmington Hills, Grosse Pointe, Royal Oak and Troy: 800-551-0488 or 248-551-1155, Option 5
- Dearborn, Taylor, Trenton and Wayne: 800-245-3725, Option 1

Laboratory Test Directory: <http://beaumontlaboratory.com/test-lab-directory>.

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**Date submitted:** December 6, 2022

**Submitted by:** Susan Daraiseh, PhD, Technical Director Molecular Pathology  
John Schwartz, MD, Medical Director Molecular Pathology