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**TO:** UCH Medical Staff, Housestaff, Patient Care Centers, and Outpatient Clinics,  
University Chicago Comprehensive Cancer Center

**FROM:** Lauren Ritterhouse, MD, PhD and Jeremy Segal, MD, PhD

**RE:** Launch of New RNA Gene Fusion Next Generation Sequencing Assay

### **Announcement**

The Clinical Genomics Laboratory in the Division of Genomic and Molecular Pathology is pleased to announce the launch of a targeted NGS assay for detection of *NTRK1*, *NTRK2*, *NTRK3*, *FGFR1*, *FGFR2*, and *FGFR3* fusion genes, available as of July 23<sup>rd</sup>, 2018.

### **Test information**

The assay is intended to detect clinically relevant *NTRK1*, *NTRK2*, *NTRK3*, *FGFR1*, *FGFR2*, and *FGFR3* fusion genes in a variety of solid tumor types, including but not limited to lung, brain, bladder and hepatobiliary tumors. Detection of these fusions may be useful for targeted therapy with anti-NTRK or anti-FGFR compounds, and can also be diagnostically useful for certain rare tumors with pathognomonic *ETV6-NTRK3* rearrangements. Acceptable specimens include formalin-fixed, paraffin-embedded (FFPE) tissue or fresh cytology smears stained via Diff-Quik. The test procedure involves RNA extraction, RNA quality/quantity assessment, RNA-seq library preparation, and targeted capture using our entire OncoPlus capture probe pool. As a result, this assay is easily extensible in the future to cover many more clinically relevant genes. After capture, samples are sequenced and the data is analyzed for detection of fusion isoforms and control gene signatures using custom-designed pipelines.

### **Specimen Requirements**

Appropriate specimens (FFPE or cytology preparations) contain >20% tumor cells and enough total cells to produce adequate RNA yield (typically >20,000 total cells). Specimens with less than 20% tumor cells may be tested at the discretion of the attending molecular pathologist.

### **Test ordering**

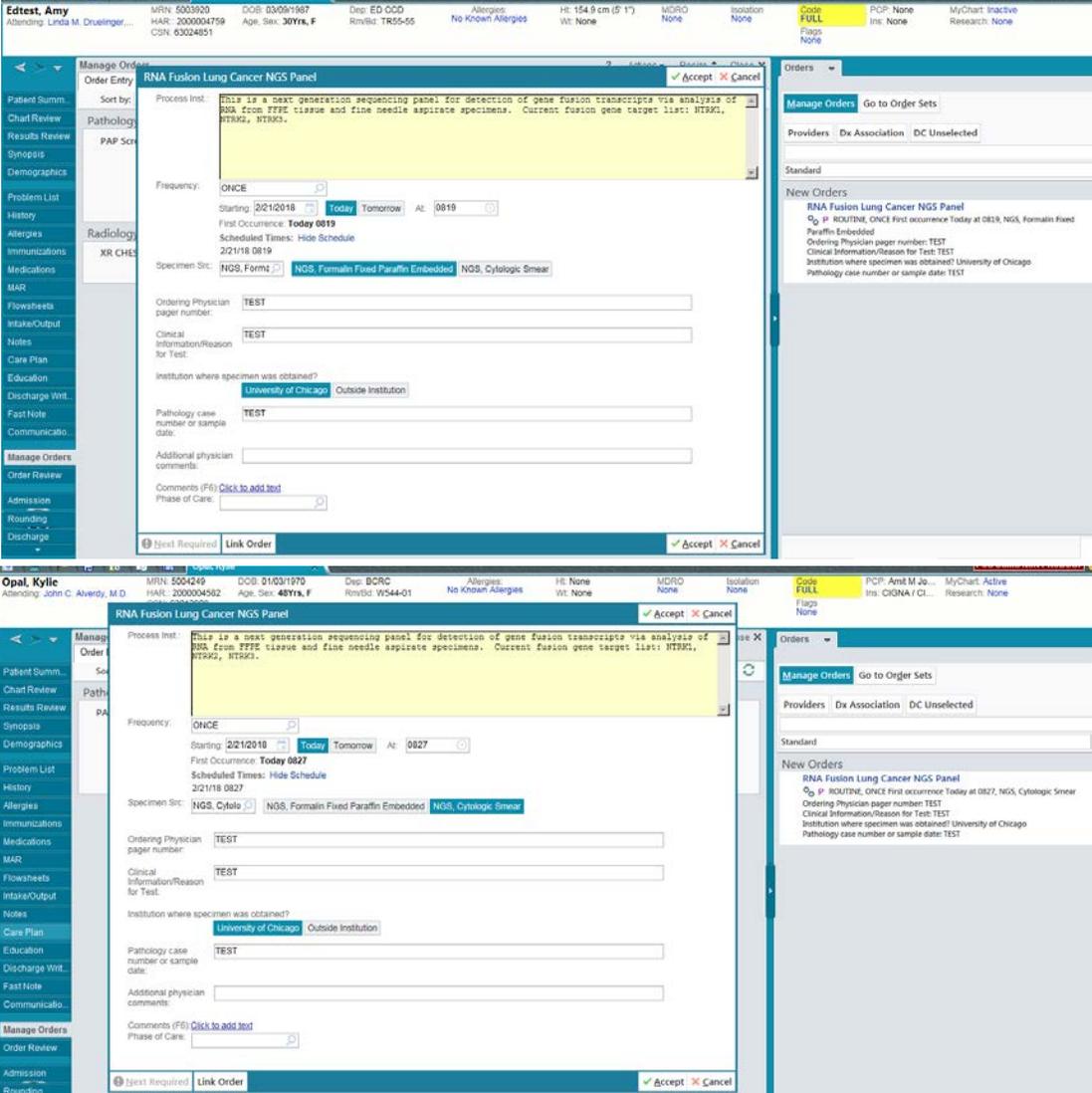
The test can be ordered through Epic using the codes LABAPFUSNFF for FFPE or LABAPFUSNCS for Diff-Quik cytology smear preparations (Figure 1).

### **Reporting and Test limitations**

The basic report format is similar to that of OncoPlus, with identified fusion genes and fusion partners reported. This test is intended for the detection of *NTRK1*, *NTRK2*, *NTRK3*, *FGFR1*, *FGFR2*, and *FGFR3* fusions in solid tumor samples with at least 20 percent tumor cells and adequate RNA. The assay has the capability to detect both previously described as well as novel fusions involving these genes. The report will contain both genes in a detected fusion pair, similar to the manner in which OncoPlus reports such fusions for *ALK*, *RET*, and *ROS1*. It should be noted that for at least the time

being, *ALK*, *RET* and *ROS1* fusions will continue to be reported as part of OncoPlus rather than as part of this new fusion assay. Mutations and copy number alterations will not be detected. Assay sensitivity is 20% tumor cell percentage, thus false-negative results may occur when there is a lower tumor cell burden.

**Figure 1.** List of Epic current order entries for the NTRK1/2/3 and FGFR1/2/3 Lung Cancer Fusion NGS Assay



**Testing Frequency and Turnaround Time**

Testing will be performed at least once weekly, Monday through Friday during day shifts only. Expected turnaround time is approximately 10-14 business days following specimen receipt.

**Additional Questions**

Additional questions may be directed to the Division of Genomic and Molecular Pathology at 773-702-4946 or Dr. Jeremy Segal at 773-702-3674 or Dr. Lauren Ritterhouse at 773-702-8491.