

Sensitive new assay finds abnormalities in tumor cells that other techniques may miss

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RNA-Seq is a new next-generation assay that can detect gene fusions in solid tumor cells with high accuracy and excellent reproducibility. According to a new report in *The Journal of Molecular Diagnostics*, the assay detected 93 percent of gene fusions identified by currently available methods with no false positives. Importantly, gene fusions missed by other techniques were found, including 18 that had never been described before. This study paves the way for clinical use of the assay to advance the diagnosis and treatment of solid tumors.

RNA-Seq is an extremely powerful [method](#) for analyzing all of the expressed genes in the genome for genetic alterations such as gene fusions, mutations, and abnormal expression that might have therapeutic, diagnostic, or prognostic significance. "It is very important to understand the accuracy, reproducibility, and sensitivity of these assays for utilities such as gene fusion detection before they are used clinically. To date very few studies have done that," explained lead investigator Kevin C. Halling, MD, Ph.D., of the Departments of Laboratory Medicine and Pathology, Division of Laboratory Genetics and Experimental Pathology at the Mayo Clinic, Rochester, MN, USA.

Using the RNA-Seq assay, researchers analyzed 76 specimens, including 61 tumor specimens and 15 normal tissues. All fusions identified were also evaluated by established diagnostic techniques such as reverse transcription-PCR and Sanger sequencing for confirmation. The test assesses for fusions in 571 genes that have been implicated in tumor formation and/or progression.

The assay was able to identify 38 of 41 gene fusions detected by other methods. No false positive results were found in normal tissues or [tumor](#) tissue that had no gene fusions present as determined by other methods (100 percent specificity). Results were found to be highly reproducible.

A particular strength of the study was its ability to detect 22 fusions in 17 tumors that had not previously been detected by another method. All 22 of these fusions were confirmed by reverse transcription-PCR. Most of these fusions (18) had never been described before, adding to the list of gene fusions associated with [solid tumors](#). Eleven of these new fusions were between gene partners that had never before been described as part of a fusion. Additional assessment will be required to determine the clinical significance of these fusions.

"Our findings make it clear that RNA-Seq has the potential to identify a large number of gene fusions that would go undetected with simpler PCR-based assays that are currently used for this purpose. This increases the likelihood that patients will receive potentially beneficial FDA-approved targeted therapies or, if such therapies are not available, will enable the patient to be entered into clinical trials for emerging therapies," noted Dr. Halling.

Gene [fusion](#) refers to the joining of two [genes](#) either by chromosomal rearrangements such as translocations or interstitial deletions. Although [gene fusions](#) were known to be common in hematologic malignancies, it has only recently been recognized that they are also common in solid tumors such as prostate and lung cancer.

More information: Development and Verification of an RNA Sequencing (RNA-Seq) Assay for the Detection of Gene Fusions in Tumors, *Journal of Molecular Diagnostics* (2018). [DOI: 10.1016/j.jmoldx.2018.03.007](#)

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