New Blood Test Capable of Detecting Multiple Types of Cancer

In study, test proved able to detect and localize more than 20 types of cancer with a high degree of accuracy.

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A new blood test in development has shown ability to screen for numerous types of cancer with a high degree of accuracy, a trial of the test shows. Dana-Farber Cancer Institute investigators [presented] the results of the multi-center trial during a session [on September 28] at the European Society for Medical Oncology (ESMO) 2019 Congress.

The test, developed by GRAIL, Inc., uses next-generation sequencing technology to probe DNA for tiny chemical tags (methylation) that influence whether genes are active or inactive. When applied to nearly 3,600 blood samples—some from patients with cancer, some from people who had not been diagnosed with cancer at the time of the blood draw —the test successfully picked up a cancer signal from the cancer patient samples, and correctly identified the tissue from where the cancer began (the tissue of origin). The test's specificity—its ability to return a positive result only when cancer is actually present—was high, as was its ability to pinpoint the organ or tissue of origin, researchers found.

The new test looks for DNA, which cancer cells shed into the bloodstream when they die. In contrast to "liquid biopsies," which detect genetic mutations or other cancer-related alterations in DNA, the technology focuses on modifications to DNA known as methyl groups. Methyl groups are chemical units that can be attached to DNA, in a process called methylation, to control which genes are "on" and which are "off." Abnormal patterns of methylation turn out to be, in many cases, more indicative of cancer—and cancer type—than mutations are. The new test zeroes in on portions of the genome where abnormal methylation patterns are found in cancer cells.

"Our previous work indicated that methylation-based assays outperform traditional DNA-sequencing approaches to detecting multiple forms of cancer in blood samples," said the study's lead author, Geoffrey Oxnard, MD, of Dana-Farber. "The results of the new study demonstrate that such assays are a feasible way of screening people for cancer."

In the study, investigators analyzed cell-free DNA (DNA that had once been confined to cells but had entered the bloodstream upon the cells' death) in 3,583 blood samples, including 1,530 from patients diagnosed with cancer and 2,053 from people without cancer. The patient samples

comprised more than 20 types of cancer, including hormone receptor-negative breast, colorectal, esophageal, gallbladder, gastric, head and neck, lung, lymphoid leukemia, multiple myeloma, ovarian and pancreatic cancer.

The overall specificity was 99.4%, meaning only 0.6% of the results incorrectly indicated that cancer was present. The sensitivity of the assay for detecting pre-specified high-mortality cancers (the percent of blood samples from these patients that tested positive for cancer) was 76%. Within this group, the sensitivity was 32% for patients with Stage I cancer; 76% for those with Stage II; 85% for Stage III; and 93% for Stage IV. Sensitivity across all cancer types was 55%, with similar increases in detection by stage. For the 97% of samples that returned a tissue of origin result, the test correctly identified the organ or tissue of origin in 89% of cases.

Detecting even a modest percent of common cancers early could translate into many patients who may be able to receive more effective treatment if the test were in wide use, Oxnard remarked.

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