

Researchers say AI blood test provides a reliable way to identify lung cancer

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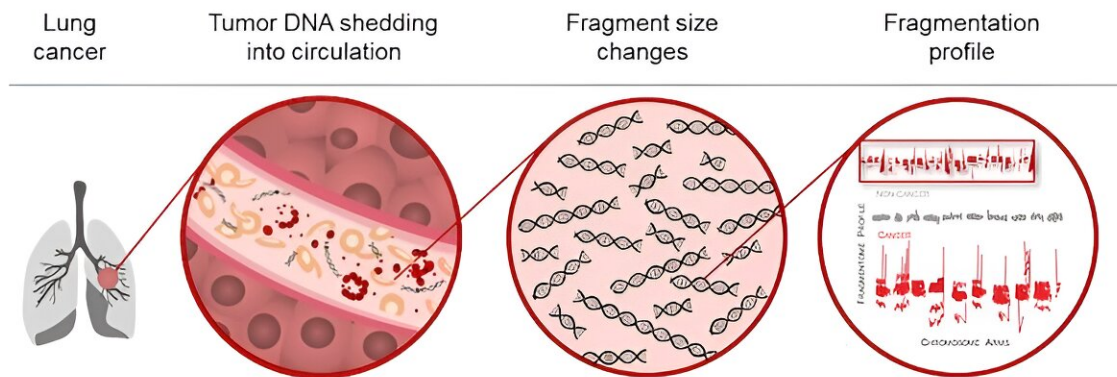


Illustration representing the DNA Evaluation of Fragments for Early Interception (DELFI) approach for lung cancer detection through non-invasive assessment of cell-free DNA fragmentation profiles. Credit: Cancer Discovery

Using artificial intelligence technology to identify patterns of DNA fragments associated with lung cancer, researchers from the Johns Hopkins Kimmel Cancer Center and other institutions have developed and validated a liquid biopsy that may help identify lung cancer earlier.

In a prospective study published June 3 in [Cancer Discovery](#), the team demonstrated that [artificial intelligence](#) technology could identify people more likely to have [lung cancer](#) based on DNA fragment patterns in the blood.

The study enrolled about 1,000 participants with and without cancer who met the criteria for traditional lung cancer screening with low-dose computed tomography (CT). Individuals were recruited to participate at 47 centers in 23 U.S. states. By helping to identify patients most at risk and who would benefit from follow-up CT screening, this new blood test could potentially boost lung cancer screening and reduce death rates, according to computer modeling by the team.

"We have a [simple blood test](#) that could be done in a doctor's office that would tell patients whether they have potential signs of lung cancer and should get a follow-up CT scan," says the study's corresponding author, Victor E. Velculescu, M.D., Ph.D., professor of oncology and co-director of the Cancer Genetics and Epigenetics program at the Johns Hopkins Kimmel Cancer Center.

Lung cancer is the deadliest cancer in the United States, according to the National Cancer Institute, and worldwide, according to the World Health Organization. Yearly screening with CT scans in high-risk patients can help detect lung cancers early, when they are most treatable, and help avert lung cancer deaths.

Screening is recommended by the U.S. Preventive Services Task Force for 15 million people nationally who are between ages 50 and 80 and have a smoking history, yet only about 6%–10% of eligible individuals are screened each year. People may be reticent to follow through on screening, Velculescu explains, due to the time it takes to arrange and go to an appointment, and the low doses of radiation they are exposed to from the scan.

To help overcome some of these hurdles, Velculescu and his colleagues developed a test over the past five years that uses artificial intelligence to detect patterns of DNA fragments found in patients with lung cancer. It takes advantage of differences in how DNA is packaged in normal and

[cancer cells](#). DNA is neatly and consistently folded up in [healthy cells](#), almost like a rolled-up ball of yarn, but DNA in cancer cells is more disorganized.

When both types of cells die, fragments of DNA end up in the blood. The DNA fragments in patients with cancer tend to be more chaotic and irregular than the DNA fragments found in individuals who do not have cancer.

The team trained artificial intelligence software to identify the specific patterns of DNA fragments seen in the blood of 576 people with or without lung cancer. Then, they verified that the method worked in a second group of 382 people with and without cancer. Based on their analyses, the test has a negative predictive value of 99.8%, meaning that only 2 in 1,000 individuals tested may be missed and have lung cancer.

The group's computer simulations showed that if the test boosted the rate of lung cancer screening to 50% within five years, it could quadruple the number of lung cancers detected and increase the proportion of cancers detected early—when they are most treatable—by about 10%. That could prevent about 14,000 cancer deaths over five years.

"The test is inexpensive and could be done at a very large scale," Velculescu says. "We believe it will make lung cancer screening more accessible and help many more people get screened. This will lead to more cancers being detected and treated early."

The test is currently available through DELFI Diagnostics for use as a laboratory-based test under the Clinical Laboratory Improvement Amendments. However, the team plans to seek approval from the U.S. Food and Drug Administration for lung cancer [screening](#). Velculescu and colleagues also plan to study whether a similar approach could be used to detect other types of cancer.

More information: Peter J. Mazzone et al, Clinical validation of a cell-free DNA fragmentome assay for augmentation of lung cancer early detection, *Cancer Discovery* (2024). [DOI: 10.1158/2159-8290.CD-24-0519](https://doi.org/10.1158/2159-8290.CD-24-0519)

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