

New blood test could reveal undiagnosed ovarian cancer in thousands of women

By Miles Martin

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Researchers have invented the first screening test for ovarian cancer that can catch the disease in its earliest stages, offering the potential to meet the urgent need for accurate, affordable ovarian cancer screening.

The test works by detecting methylated DNA, regions of genetic material that have been modified with chemical markers. While DNA methylation has been studied in relation to ovarian cancer, this is the first time that this connection has been applied to screen for this deadly disease. The [patent application](#) for the invention was published May 6 by the U.S. Patent and Trademark Office.

"Early detection or screening is a challenging word to use when it comes to [ovarian cancer](#), because there's really no good screening test for ovarian cancer," said Jamie Bakkum-Gamez, a gynecologic oncologist and a professor of obstetrics and gynecology at the Mayo Clinic in Rochester, Minnesota. Bakkum-Gamez served as the principal investigator for this invention, as well as for related inventions, which are at earlier stages in the patent

process and are intended for screening against cervical and uterine cancer.

The [American Cancer Society](#) estimates that 21,410 women in the United States will receive a new diagnosis of ovarian cancer in 2021, and about 13,770 will die from the disease. Ovarian cancer accounts for more deaths than any other cancer of the female reproductive system, ranking fifth overall in cancer deaths among women. And though 94% of patients live longer than five years after diagnosis when [ovarian cancer](#) is caught in its earliest stages, only about 20% of cases are identified early.

The U.S. Preventive Services Task Force, an independent panel of experts that provides recommendations for preventive medicine, goes so far as to recommend that women not be screened for [ovarian cancer](#) if they do not have symptoms, because frequent false positives from current methods can lead to unnecessary surgery for women without cancer.

But waiting for symptoms is not ideal, because many of the earlier symptoms of ovarian cancer, such as abdominal distention, bladder changes or even just feeling full, have given ovarian cancer a reputation for being, as Bakkum-Gamez put it, a "silent killer."

"The problem is that those symptoms can be present in early-stage cancer or advanced-stage cancer — or [the women] can be pregnant," she said. "They can have zero symptoms and have an ovarian cancer brewing."

To create a better test, the researchers sought to zero in on a marker of ovarian cancer that, unlike the early symptoms, is specific enough to the cancer to indicate its presence. They found the answer in epigenetics, a field that studies changes that occur in DNA that do not alter the original DNA sequence. Bakkum-Gamez said that epigenetics has already been employed in screening against [gastrointestinal cancer](#), where it has proved more efficient at detecting cancer at earlier stages.

Epigenetic changes occur in response to a variety of factors, such as diet and lifestyle choice. [Epigenetics](#) may explain why some diseases, including ovarian cancer, are known to run in families but do not appear to be passed down through specific genes. The researchers focused on DNA methylation, one of the most well-known epigenetic markers.

"Methylation is an epigenetic change on the DNA that usually happens at the promoter, which is the part that's the on-off switch for the gene," Bakkum-Gamez said.

"Usually, these types of genes are tumor-suppressor genes, which are the genes that are preventing our cells from developing into cancer."

The test works by isolating methylated DNA in these tumor-suppression genes and then amplifying it to levels where it can be detected in a liquid biopsy. And while the test can be done on ovarian tissue directly, it can also be done on blood, making it much less invasive than a full

biopsy, because many women can easily collect blood from tampons.

"[It's] somewhat inspired by the success of Cologuard," Bakkum-Gamez said, referring to an at-home colon cancer test that uses a stool sample from the patient, avoiding a trip to the clinic.

In an initial study published in 2020, the researchers validated the feasibility of using these markers to test for ovarian cancer in 122 tissue samples and are currently working on another study on a larger sample size. So while the inventors are optimistic about their test, it may be some time before it is available to patients.

"I would have to say that we're still in the very exciting but early stages," Bakkum-Gamez said.

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The researchers hope that, in the future, their invention will see widespread application for women all over the world, particularly those who may be predisposed to ovarian cancer because of family history.

"I love science, but I am a clinician, and so I want this to be something that makes a difference for my patients and for their offspring, because some of our patients actually develop ovarian cancer because of a genetic condition," Bakkum-Gamez said. "And so I want something better out there for our patients who are at high risk for ovarian cancer."

The application for the patent, "Detecting ovarian cancer," was filed with the U.S. Patent and Trademark Office on Oct. 30, 2020. It was published May 6 with the application number 2021/0130907. The earliest priority date was Oct. 31, 2019. The inventors of the pending patent are William R. Taylor, John B. Kiesel, Douglas W. Mahoney and David A. Ahlquist, Mayo Clinic; and Hatim T. Allawi and Michael W. Kaiser, Exact Sciences.