

Intermountain healthcare researchers launch major three-year genomics breast cancer study

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News Intermountain Healthcare study is unique in that researchers will also help develop a specific test to check for ctDNA, and will have access to both mammography results and the DNA blood test results, which will allow a direct comparison of the 'liquid-based biopsy' to be made. Credit: Intermountain Medical Center

Cancer researchers at Intermountain Medical Center and the Intermountain Precision Genomics Program in Salt Lake City are launching an exciting, new three-year study to determine if a blood test that looks for DNA from a cancer tumor can be used to complement mammography to improve the way breast cancer is diagnosed.

The goal of this new genomics study is to show whether screening patients for the presence of circulating tumor DNA, known as ctDNA, can successfully detect [breast cancer](#) using a blood draw.

Breast cancer is the second-leading cause of cancer deaths in women, behind only lung cancer, with an estimated 40,610 deaths each year from the disease. Nearly 253,000 new cases of [invasive breast cancer](#) are diagnosed each year, along with about 60,000 non-invasive, early-stage cases, according to the American Cancer Society.

The Intermountain study is unique in that researchers will also help develop a specific test to check for ctDNA, and will have access to both [mammography](#) results and the DNA [blood test](#) results, which will allow a direct comparison of the "liquid-based biopsy" to be made.

The idea behind the science is simple, though researchers say the execution is not yet proven: Little pieces of DNA that come from dying cells end up in the peripheral blood stream, including circulating tumor cells. The goal of researchers is to use those markers to identify breast cancer, perhaps even before mammography can detect it, said Lincoln Nadauld, MD, PhD, co-lead investigator of the study and executive director of the Intermountain Healthcare Precision Genomics Program.

"As a tumor is growing, some of the cells will die and their DNA will end up in the peripheral blood stream," Dr. Nadauld said. "We're able to distinguish DNA from cancer vs. DNA from normal cells. The idea is to leverage DNA to see if we can detect that it comes from a tumor."

In the study, patients with known breast cancer will be compared with those in a screening group.

"We don't know what we'll see yet," said Brett Parkinson, MD, co-lead investigator of the study, who is also imaging director and medical director of the Intermountain Medical Center Breast Care Center in Murray. "We might find those who have breast cancer will have a negative blood test and learn it's not a good screening tool."

Even a successful blood test isn't expected to replace mammography outright. If it detects the circulating tumor DNA, imaging would be needed to find the tumor. But it could help eliminate unneeded biopsies, Dr. Parkinson added.

Dr. Nadauld said cancers have mutations in their DNA that aren't always unique.

"Sometimes those are the same whether it's a breast cancer or a colon cancer. If we do create a blood test, it's possible it would detect mutant DNA, but it might look so similar it would be hard to tell what kind of cancer it came from," he said. "That's part of what this trial is going to accomplish. We want to determine the signature for early breast cancer."

If successful, a liquid biopsy might also be used to monitor a breast cancer survivor for recurrence, Dr. Nadauld said. It might even lead to development of similar tests for different types of cancer. But that would be a challenge for the future.

"We want to approach this with laser-like focus," he said. "It's needed to help us diagnose breast cancer. We need to detect it earlier, when it's curable."

Breast cancer survival depends largely on finding the disease early —and

mammography is the only screening exam that's been shown by multiple randomized clinical trials to reduce the mortality rate for breast cancer. Since 1991, the death rate from breast cancer is down 38 percent, largely because mammography screening tests lead to early detection.

Although mammography finds most breast cancers, it may not detect malignancy in women who have dense [breast tissue](#), especially premenopausal women, or those under 50.

"We pick up most breast cancer in women with average breast density," said Dr. Parkinson. "When breast tissue is denser, we can miss up to 30 percent of breast cancers."

Mammography also has a false-positive or call-back rate of 10 percent, which may subject women to additional imaging and emotional duress. Plus, a mammogram can be uncomfortable, since breast tissue is compressed for imaging, which also exposes a woman to a small amount of radiation. Mammography may also be inconvenient, often requiring women to take time off work, he noted.

For those, and perhaps other reasons, mammography screening rates in the United States are low. In Utah, only about 65 percent of eligible women are screened, despite Intermountain Healthcare's recommendations that women over 40 undergo yearly screening mammography. All major medical and advocacy organizations agree that screening every year after a woman is 40 saves more lives. About 20 percent of breast cancers occur in women under 50.

Dr. Nadauld said the unusual confluence of three factors weigh in Intermountain's favor on this quest, starting with access to a lot of patients in one place who are getting mammograms, which are the gold standard screening test for breast cancer. Second, the researchers have access to the results of those mammograms; they know if the results

were positive or negative. The third major factor is Intermountain's genomic technology capability.

"This is the big conversation right now in all of oncology — the use of liquid biopsy to determine how to screen for [breast cancer](#), a woman's risk of recurrence, and how to monitor their treatment," Dr. Nadauld said.

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Provided by Intermountain Medical Center

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