

Study confirms cell-free DNA in urine as potential method for cancer detection

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Urinalysis has long been a staple of physical exams to detect and manage a number of diseases and disorders, but not cancer. What if it were that easy, though, and cancer was detected in its very earliest stages when the disease responds more favorably to treatment and improved outcomes are more likely?

That was the question posed by scientists at the Translational Genomics Research Institute (TGen), an affiliate of City of Hope, who have found a way of zeroing in on early-stage [cancer](#) by analyzing short strands of cell-free DNA in urine. Their study's findings were published today in the scientific journal *Science Translational Medicine*.

Previous thought once held that DNA fragments in urine were degraded at random and were too short to provide any meaningful information about a disease as complex as cancer. TGen and City of Hope researchers and their colleagues from Baylor University and Phoenix Children's Hospital found that these DNA fragments are not random at all, and can clearly indicate a difference between healthy individuals and those with cancer.

"There are many steps between where we are now and where we want to go—detecting cancer from a urine sample—but without doubt this is an encouraging first step," said Muhammed Murtaza, M.B.B.S., Ph.D., an Associate Professor and Co-Director of TGen's Center for Noninvasive Diagnostics, and the study's senior author.

Dr. Murtaza previously led a team of TGen scientists who pioneered the use of circulating tumor DNA in blood, using genetic fragments to detect cancer with a simple blood draw. This "liquid biopsy" method sidesteps the need for many surgical biopsies of suspected tumors, and means that physicians can monitor cancer in their patients more frequently given the less invasive nature of the procedure.

Collecting a [urine sample](#) reduces the physical invasiveness to zero, Dr. Murtaza explained, and may eliminate a lab visit, given that the sample could be collected at home and mailed in for analysis.

By studying tissue samples from children with various cancers, whose malignancies often move extraordinarily fast, and adults with pancreatic cancer, whose early detection is critical to their disease outcomes, researchers mapped the DNA fragmentation profiles in their urine.

"We found that certain regions of the genome are protected from fragmentation in urine from healthy individuals, but the same regions are more fragmented in patients with cancer," Dr. Murtaza said.

The fragmentation profiles were remarkably similar across multiple individuals; the length of the DNA fragments were similar, the regions of the genome where the fragmentation occurred were consistent, and informed researchers what type of cells contributed the fragments.

Ajay Goel, Ph.D., chair of the Department of Molecular Diagnostics and Experimental Therapeutics and Associate Director for Basic Science at City of Hope, a world-renowned independent research and treatment center for cancer and diabetes, is one of the study's authors. He is a leading expert in developing early-detection blood tests for colon, pancreatic and ovarian cancers.

"If the study results come to fruition, our urinalysis technology would be a remarkable breakthrough in the detection of many cancers, especially in pancreatic cancer," Dr. Goel said. "If cancer is detected early, it could substantially lower the mortality rate for what is currently the third leading cause of cancer death in the U.S."

While early results are promising, the researchers indicate the need to test their findings in much larger populations of cancer patients and identify differences between men and women, young and old, and those with co-morbidities, such as diabetes and other chronic diseases.

"This is a fundamental new finding and provides a potentially dynamic path forward for the early diagnosis of cancer, given that [urine](#) is one of the easiest samples to collect," said Daniel D. Von Hoff, M.D., TGen Distinguished Professor and one of the paper's authors. "If follow-on studies yield positive results, I could one day see this test becoming an integral part of one's annual physical."

More information: Havell Markus et al, Analysis of recurrently protected genomic regions in cell-free DNA found in urine, *Science Translational Medicine* (2021). [DOI: 10.1126/scitranslmed.aaz3088](https://doi.org/10.1126/scitranslmed.aaz3088)

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