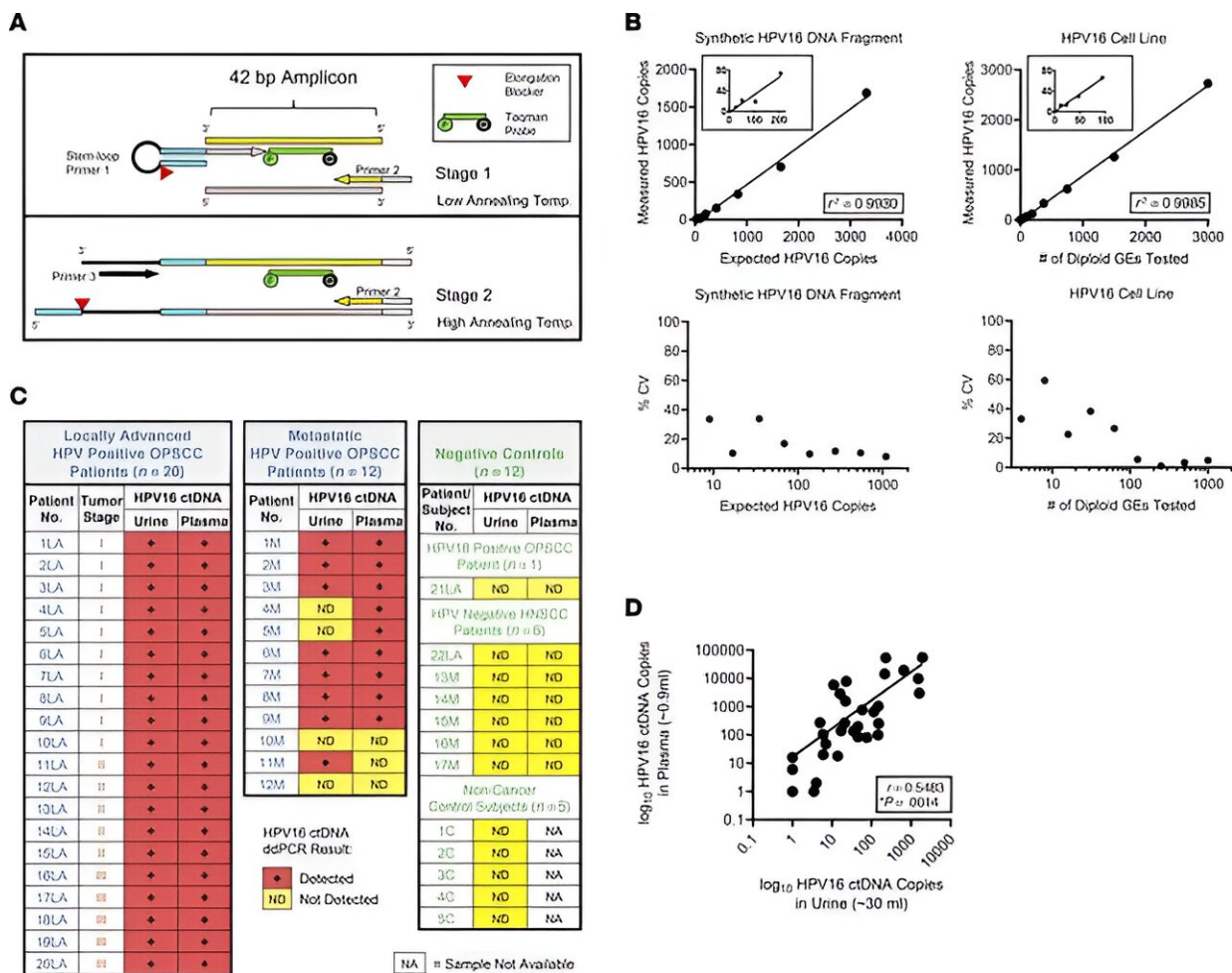


# A urine-based test that detects tumor DNA fragments could offer early reliable screening for head and neck cancer

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Ultrashort amplicon HPV16 stem-loop ddPCR assay detects TR-ctDNA fragments in urine from patients with HPV+ OPSCC and shows concordance with results from plasma ctDNA analysis. Credit: *JCI Insight* (2024). DOI:

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Researchers from the University of Michigan Health Rogel Cancer Center have created a urine-based test that detects pieces of DNA fragments released by head and neck tumors. The test could potentially facilitate early detection of this cancer type, which currently does not have a reliable screening method.

Human papillomavirus (HPV) is widely recognized for causing [cervical cancer](#), but is increasingly found to cause cancers in the mouth, throat and other head and neck regions.

Early detection is critical because detecting a cancer at an earlier stage can lead to better outcomes for patients.

Using [whole genome sequencing](#), the Rogel group showed that cell-free DNA fragments released by tumor cells, which are passed on from the bloodstream into urine through the kidneys, are predominantly ultrashort, with fewer than 50 base pairs. Given their small size, these fragments are likely to be missed using conventional urine or blood-based liquid biopsy tests in detecting circulating tumor DNA (ctDNA).

The research was led by Muneesh Tewari, M.D., Ph.D., professor of hematology and oncology, J. Chad Brenner, Ph.D., associate professor of otolaryngology-head and neck surgery, and Paul L. Swiecicki, M.D., associate medical director for the Oncology Clinical Trials Support Unit at Rogel. Initial results are [published](#) in *JCI Insight*.

"In this study we provide evidence to support the hypothesis that conventional assays do not detect ultrashort fragments found in urine, since they are designed to target longer DNA fragments. Our team used

an unconventional approach to develop a urine test for HPV-positive head and neck cancer ctDNA detection," said study co-first author and research specialist Chandan Bhambhani, Ph.D.

Still in the discovery phase, this mail-in test has already been distributed for research purposes to patients within a hundred-plus miles from Ann Arbor, allowing scientists to gather significant data on the efficacy of the at-home kit. Participants collect a [urine sample](#) and have it shipped back to the U-M laboratory, where the testing can be done to detect the presence or absence of head and neck cancer.

"One of the most remarkable outcomes of this study is that the test that has been developed has detected cancer recurrences far earlier than would typically happen based on clinical imaging. As such, these promising results have given us the confidence to broaden the scope of the study, seeking to expanding distribution even further," said Brenner, co-senior author of the study.

While initial studies have focused on head and neck cancer, the paper also describes a new method that could be applied to expand the test to detect other cancers as well. For example, the authors show that the test can detect ctDNA in the urine of patients with breast cancer and acute myeloid leukemia. This suggests new opportunities to also study the application of urine-based testing for these additional cancers.

"Many people are not aware that urine carries information about many different cancer types, although it is made in the kidneys. Our findings about the difference in ctDNA fragment sizes and the test we developed for HPV-positive head and neck cancer detection provide crucial information on how urine-based diagnostic assays can be developed for different cancers," Bhambhani said.

"Further, these types of tests are likely to have a much higher

compliance in patients requiring follow-up testing post treatment, due to the convenience of self-collection of samples, when compared to blood-based assays."

**More information:** Chandan Bhambhani et al, ctDNA transiting into urine is ultrashort and facilitates noninvasive liquid biopsy of HPV+ oropharyngeal cancer, *JCI Insight* (2024). [DOI: 10.1172/jci.insight.177759](https://doi.org/10.1172/jci.insight.177759)

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