

Promising urine DNA test could facilitate detection of urothelial cancer

Lyon, France, 20 May 2019 – Scientists at the International Agency for Research on Cancer (IARC), in collaboration with researchers at the Protestant Clinic of Lyon (France), the Portuguese Oncology Institute of Porto (Portugal), and Lomonosov Moscow State University (Russian Federation), have developed a simple, sensitive, and specific urine test (called UroMuTERT) that is capable of detecting urothelial cancer (carcinoma of the bladder and of the upper urinary tract).

These exciting new results were published today in an article in *EBioMedicine*,¹ a journal published by *The Lancet*. The new test could profoundly change the way in which urothelial cancer is detected and clinically managed.

“This non-invasive test could be very beneficial for patients,” says Dr Florence Le Calvez-Kelm, a scientist in the Genetic Cancer Susceptibility Group at IARC and the principal investigator of the study. “We are still at an early stage in the development of biomarkers for the detection of urothelial cancer, but this test has the potential to significantly improve early detection and reduce the number of unnecessary invasive procedures, such as cystoscopy.”

The test builds on the discovery of two highly recurrent mutations in the telomerase reverse transcriptase (*TERT*) gene; these mutations are present in any kind of urothelial tumour and can be detected in cells shed into the urine of patients with urothelial cancer. The UroMuTERT test was developed specifically for the detection of low levels of *TERT* mutations in bodily fluids.

Despite many previous attempts to improve the clinical management of urothelial cancer through the use of so-called liquid biopsies, the existing urinary biomarkers are suboptimal. Some are not specific enough, some are not cost-effective, and some do not detect all forms of urothelial cancer. Therefore, the detection and monitoring of urothelial cancer still relies largely on a combination of cystoscopy, which is invasive and expensive, and urine cytology, which is non-invasive but fails to identify most low-grade lesions.

The researchers evaluated the clinical performance of UroMuTERT for the non-invasive detection of urothelial cancer in 93 patients with a confirmed diagnosis (cases) and 94 individuals without the disease (controls) in France and in 50 cases and 50 controls in Portugal.

The UroMuTERT test demonstrated excellent sensitivity and specificity for the detection of all forms of urothelial cancer. The test, which detects *TERT* mutations in DNA in urine, was found to significantly outperform urine cytology for the detection of low-grade early-stage tumours.

¹ Avogbe PH, Manel A, Vian E, Durand G, Forey N, Voegelé C, et al. (2019). Urinary *TERT* promoter mutations as non-invasive biomarkers for the comprehensive detection of urothelial cancer. *EBioMedicine*. Published online 20 May 2019; <https://doi.org/10.1016/j.ebiom.2019.05.004>

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These results indicate that the UroMuTERT urine DNA test has the required clinical performance to detect urothelial cancer at diagnosis. Researchers are currently assessing the ability of the test to detect urothelial cancer before clinical diagnosis.

In the article, the researchers also propose a strategy for integrating the detection of *TERT* mutations in DNA in urine as a primary diagnostic tool for individuals who are at high risk of developing urothelial cancer or are under surveillance for recurrence of urothelial cancer.

“In addition to its high sensitivity and specificity, the test provides a simple and inexpensive tool to detect the disease and could be a real asset to urological experts to consider its implementation in current medical standards,” says IARC scientist Dr Patrice Avogbe, the first author of the study.

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