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Considering genetic risk in prostate cancer referrals could lead to earlier diagnosis

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Men at the highest risk for prostate cancer could be fast-tracked for investigation if their genetic risk was considered in general practice, new research has concluded.

A large-scale study by the University of Exeter, published in the *British Journal of Cancer*, looked at the impact of incorporating genetic risk for cancer into the GP triage and referral processes. The research concluded that considering genetic risk could improve referrals for those in need—and importantly, avoid invasive biopsy investigations for those at low risk of cancer. Assessing genetic risk in primary care could lead to earlier diagnosis for men most at risk of prostate cancer.

Prostate cancer accounts for around a quarter of new cancer cases in men—approximately 52,000 men are diagnosed per year in the UK alone. It is the second most common cause of cancer death in men in the UK, and five-year survival doubles if it's diagnosed at an early stage compared to advanced stage. Symptoms are common and easily misdiagnosed, and an estimated 14 percent of prostate cancer deaths could be avoided if they were diagnosed earlier.

GPs make around 800,000 suspected prostate cancer referrals annually in the UK. The research team estimate that incorporating genetic risk for cancer into GP triage could mean 160,000 men could be expedited for faster investigation, while 320,000 of these could safely avoid referral and unpleasant investigation.

Lead author Dr. Harry Green, independent Research Fellow at the University of Exeter Medical School, said: "Our study is the first to demonstrate that incorporating genetic risk into GP's risk assessment of patients' symptoms of possible prostate cancer could result in faster referral for those at most risk."

At the moment, a prostate Specific Antigen (PSA) test is used to investigate men with erectile dysfunction or urination problems, but the accuracy of the test is unclear, and false positive results are common. Only one in three men with a positive PSA test have cancer. An invasive and unpleasant biopsy is often needed for diagnosis. Research shows the

PSA test can miss around 15 percent of cancers.

The team calculated genetic risk for prostate cancer using more than 250 known genetic variants linked to the disease. These genetic variants are combined into a single 'genetic risk score' which describes an individual's genetic risk of developing prostate cancer. They applied this to data from 6,390 White European men from UK Biobank, whose records showed they had seen a GP with potential prostate cancer symptoms.

The study's lead investigator, Dr. Sarah Bailey, Senior Research Fellow at the University of Exeter Medical School, said: "This is potentially an exciting new strategy for early cancer detection. Not only can high risk patients be fast tracked, but those at low risk can safely avoid invasive investigations. Using this technique would align well to the NHS Long Term Plan, which pledges to become the first national health care system to offer whole genome sequencing as part of routine care. This could be a clear example of improving early diagnosis, and therefore treatment and survival".

Kirsten Higgins, whose family are long-term supporters of the University of Exeter, funded the study. She said: "We're delighted to be able to support the Exeter team to explore the application of genomics data in a more targeted approach to prostate cancer detection. It's very exciting to see the real world benefit to patients of this innovative new approach."

Months of anxiety and testing

Richard Westlake endured 18 months of anxiety before he was declared clear of prostate cancer. Now 74, Mr Westlake, a retired Devon County Councillor and railway train driver, was first alerted in November 2015 that his night-time trips to the toilet may be a sign of the condition, combined with a raised PSA test.

Mr Westlake, from Exeter, then faced months of hospital visits, first for a biopsy, then for a more invasive biopsy procedure, along with X-rays and a combination of other tests, each followed by an anguished wait for the result. Finally, after a second biopsy in June 2017, he was given the all-clear. "I was quite surprised to be honest," he said of the result. "I'd been expecting it to be positive, all that time. But I wasn't celebrating—I'd been through a very difficult time, with all that testing, waiting, unpleasantness and anxiety. I knew it had to be done, and all the staff I met were brilliant, but it was a horrible, drawn-out experience.

"Prostate cancer is one of the most common cancers in men, and you think of all the hours of NHS time and resource going into this testing. If this new way of assessing patients could cut down on the number of men who have to experience that, I think it would have huge benefits, both to patients and to the NHS."

The study is entitled 'Applying a genetic risk score for prostate cancer to 2 men with lower urinary tract symptoms in primary 3 care to predict prostate cancer diagnosis: a cohort 4 study in the UK Biobank', and is published in the *British Journal of Cancer.*

More information: Harry D. Green et al, Applying a genetic risk score for prostate cancer to men with lower urinary tract symptoms in primary care to predict prostate cancer diagnosis: a cohort study in the UK Biobank, *British Journal of Cancer* (2022). DOI: 10.1038/s41416-022-01918-z

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