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Genetic test would help 'cut bowel cancer spread'

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Screening families of patients with bowel cancer for a genetic condition would cut their risk of developing bowel, womb, and ovarian cancers, new research has found.

In a major study, Dr Ian Frayling from Cardiff University's School of Medicine and researchers from the University of Exeter's Medical School assessed the effectiveness of introducing a UK-wide screening programme for a genetic condition known as Lynch Syndrome.

Lynch syndrome is caused by changes in genes which check the spelling in DNA. The condition increases the risk of people developing cancer, particularly bowel cancer and cancers of the womb and ovaries later in life. Without testing cancers, it is not obvious that they are caused by Lynch syndrome, and so it is often not diagnosed.

It is responsible for around one in 12 cases of people aged under 50 and around a third of people with the disease develop bowel cancer by the time they are 70, if no action is taken.

"If Lynch Syndrome is identified as the cause of bowel cancer, patients can be offered risk-reducing measures such as more intensive post-operative colonoscopy surveillance to spot recurrences and new cancers early," according to Cardiff University's Dr Ian Frayling, the clinical adviser to the study.

"As close relatives have a 50 per cent chance of sharing the gene,

screening would provide a valuable opportunity to detect the condition in children, siblings, parents and more distant relatives.

"It would mean measures could be taken to reduce the risk of cancers developing," he added.

The findings, published in Health Technology Assessment, indicate that screening the 1,700 people under the age of 50 who are newly diagnosed with bowel cancer (in England) each year would identify two thirds of these whose cancer was caused by Lynch Syndrome.

From this group, the findings suggest that 40 further cases of cancer could be avoided in them and their relatives.

Dr Frayling adds: "This is a very significant piece of work which is much to be welcomed.

"It justifies the National Health Services in the UK in implementing such testing, which is already carried out in other European countries.

"Those with Lynch Syndrome will now be found and given the care that they warrant, saving time, lives, money and resources. Colleagues around the world are eager to use the model developed by the University of Exeter's Medical School, so the benefits extend far beyond the UK."

The team systematically reviewed all the evidence surrounding Lynch Syndrome and bowel cancer, identifying and assessing 42 studies in total, before constructing a computer model of screening strategies for Lynch Syndrome.

It found that all screening strategies helped improve health outcomes at a cost generally acceptable to the NHS. The most cost-effective method of identifying Lynch Syndrome involved running tests on the tumour before

offering counselling and genetic testing.

In a separate analysis, which has not yet been published or peer-reviewed, the team used the same model to estimate that 28 cancer related deaths (24 from bowel cancer and four from womb cancer) could be prevented each year if Lynch Syndrome screening for people with bowel cancer was introduced.

Dr Tristan Snowsill, of the University of Exeter's Medical School, said: "This is the first evidence that systematic testing for Lynch syndrome could be cost-effective in the NHS.

"There are health professionals in the NHS who think cost-effectiveness is the hurdle that needs to be cleared before systematic testing can be implemented; policymakers now have that evidence before them to decide if this is right for the NHS, a decision which will not be solely based on cost-effectiveness."

Deborah Alsina, Chief Executive of Bowel Cancer UK, commented: "We welcome these research findings which demonstrate that lives can be saved through earlier identification of those who are at higher risk of bowel cancer because of genetic conditions like Lynch syndrome.

"It's critical that more lives are saved by ensuring people gain access to the screening surveillance they need, so that bowel cancer can be ruled out first, not last, in younger patients.

"While bowel cancer is thankfully relatively rare in people under 50, there are still 550 people in this age group who lose their lives to bowel cancer each year and that must change."

The research was funded by the National Institute for Health Research (NIHR), with support to Dr Frayling from the National Institute for

Social Care and Health Research (NISCHR) via Cardiff and Vale University Health Board.

More information: A systematic review and economic evaluation of diagnostic strategies for Lynch syndrome, Snowsill T, Huxley N, Hoyle M, Jones-Hughes T, Coelho H, Cooper C, Frayling I, Hyde C, *Health Technology Assessment* Volume: 18 Issue: 58, September 2014. DOI: 10.3310/hta18580

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