New genetic screening test for Lynch Syndrome

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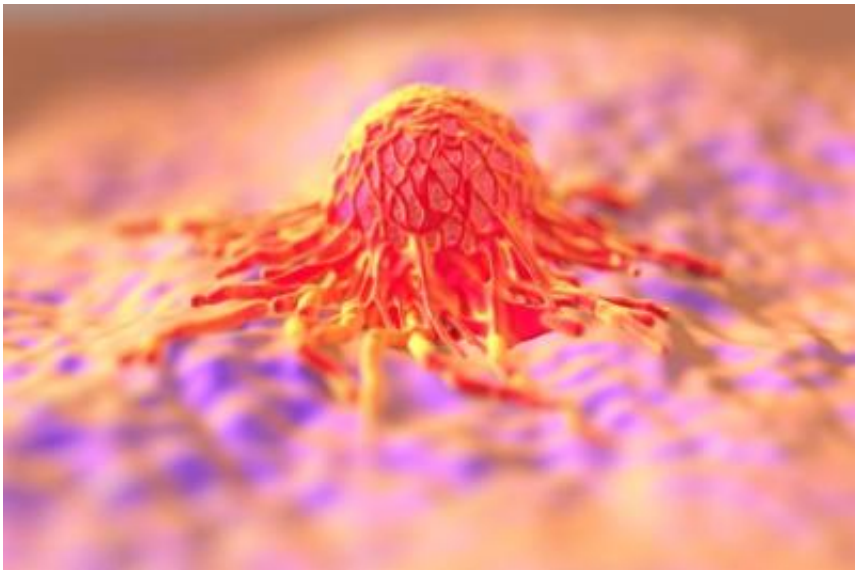

People at high risk of cancer of the bowel and womb due to a genetic condition will receive a more accurate diagnosis as a result of a new model developed by a team of international scientists.

A scientist from the University's Institute of Medical Genetics, School of Medicine, is part of an international research team who have developed this new way of identifying people at high risk of cancer.

The research, published in the journal *Nature Genetics*, focuses on the

genes responsible for Lynch Syndrome - an inherited, familial condition that increases the risk of bowel, womb and other cancers, especially at a young age. About 5% of bowel and womb cancers are thought to be due to Lynch Syndrome.

The model essentially turns previously uninterpretable DNA data into usable knowledge with a direct clinical benefit. It means doctors worldwide can now access new information and give patients a truer picture of their familial risk.

"In the UK, bowel cancer kills about 16,000 people each year, and womb cancer - the commonest gynaecological cancer - about 2,000 women a year, with Wales having the highest rate," according to Dr Ian Frayling, School of Medicine, member of the International Society for Gastrointestinal Hereditary Tumours (InSiGHT) which organised the research.

"What we have been able to do is effectively refine genetic information in the InSiGHT database and provide a more accurate answer of the risk of getting cancer," he added.

Patients who currently have genetic testing for Lynch Syndrome are often told they have 'variants of uncertain significance' which is an inconclusive result.

People are left in a kind of genetic limbo, unsure of whether they face a high risk of cancer, or whether their family members are also at risk.

"As a result of this work, doctors will now be able to say much more confidently whether those patients have Lynch Syndrome, and therefore whether they are at a higher risk of cancer," according to Dr Frayling.

"This will help to save more lives, because by giving a definite answer to

more patients, they will be able to access the specialist screening that they need.

"It will also save NHS resources and be safer, because we will be able to reassure people that they are not at risk and so don't need the extra screening. As a result of this, colleagues and the families they care for all over the UK are now benefitting, and we are working on incorporating this into the UK Guidelines on testing for Lynch Syndrome," he adds.

Provided by Cardiff University