Identification of 'fingerprint' of rare tumor leads to development of cheap and reliable new test

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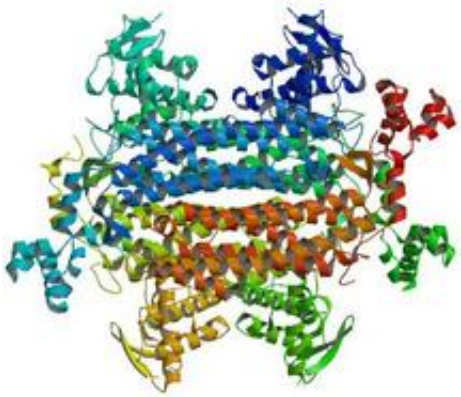

A new test for a rare form of cancer picks a biological ‘fingerprint’ of a deficiency in the FH enzyme.

Researchers at the University of Oxford have developed a cheap and reliable diagnostic test for a rare form of cancer. The test involves screening tumour samples for a particular molecular fingerprint unique to this type of cancer.

Hereditary leiomyomatosis and renal cell cancer (HLRCC) is a disorder which causes the development of benign but often painful tumours in the skin and, in females, in the uterus. Between one in six and one in ten people affected by the disorder will go on to develop an aggressive form of kidney cancer called papillary renal cell cancer. The condition often

strikes people in their twenties.

The disorder is caused by mutations, which may be inherited, in a gene responsible for the production of an enzyme known as fumarate hydratase (FH). This leads to an accumulation within cells of fumarate, which promotes the development of cancer cells.

Now, in a study published in The Journal of Pathology, an international team of scientists led by researchers at the Henry Wellcome Building for Molecular Physiology, University of Oxford, have identified a particular protein modification which is induced by FH deficiency (and hence an over-abundance of fumarate). This alteration is unique to this type of tumour and can hence be used as a biomarker – a biological 'fingerprint' to identify tumours caused by this mechanism.

The researchers have developed a test for this protein modification which can be carried out in less than two hours and will identify tumours with FH mutations. This approach is much more cost effective than genetic testing of all possible cases using DNA sequencing. They show that screening cases of papillary renal cell cancer using this new test allows them to identify undiagnosed cases of HLRCC for genetic testing. They believe this test should be applied to all cases of papillary renal cell cancer to identify those with FH mutations, allowing advice to be provided to their families on their own relative risks of developing the disorder and associated kidney cancer.

"Cancer can be caused by many different risk factors, but if we can pinpoint rapidly and accurately the particular type of tumour, we can provide more accurate advice to patients and their families, and perhaps diagnose cases at earlier, more treatable, stages," explains Dr Patrick Pollard, a Beit Memorial Fellow at the University of Oxford. "For the first time, we are now able to screen for tumours caused by this rare, but often very serious, condition using a test which is simple, cheap and

reliable."

Dr Pollard and colleagues have filed a patent to develop the test, which is currently being marketed by Isis Innovation at the University of Oxford.

Dr Lesley Walker, director of cancer information at Cancer Research UK, said: "We know that diagnosing cancer at an earlier stage offers the best chances of successful treatment. So being able to identify other family members who are at risk so they can be monitored more closely is crucial to improving survival rates from this rare aggressive form of kidney cancer.

"Tests like this can also help us to identify other patients with the same mutation, paving the way for the development of targeted treatments for specific groups of patients. This approach is called stratified medicine and many scientists now believe it could revolutionalise cancer treatment in the future."

Provided by Wellcome Trust

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