Medical Xpress

Low cost, safe and accurate test could help diagnose rare childhood cancers

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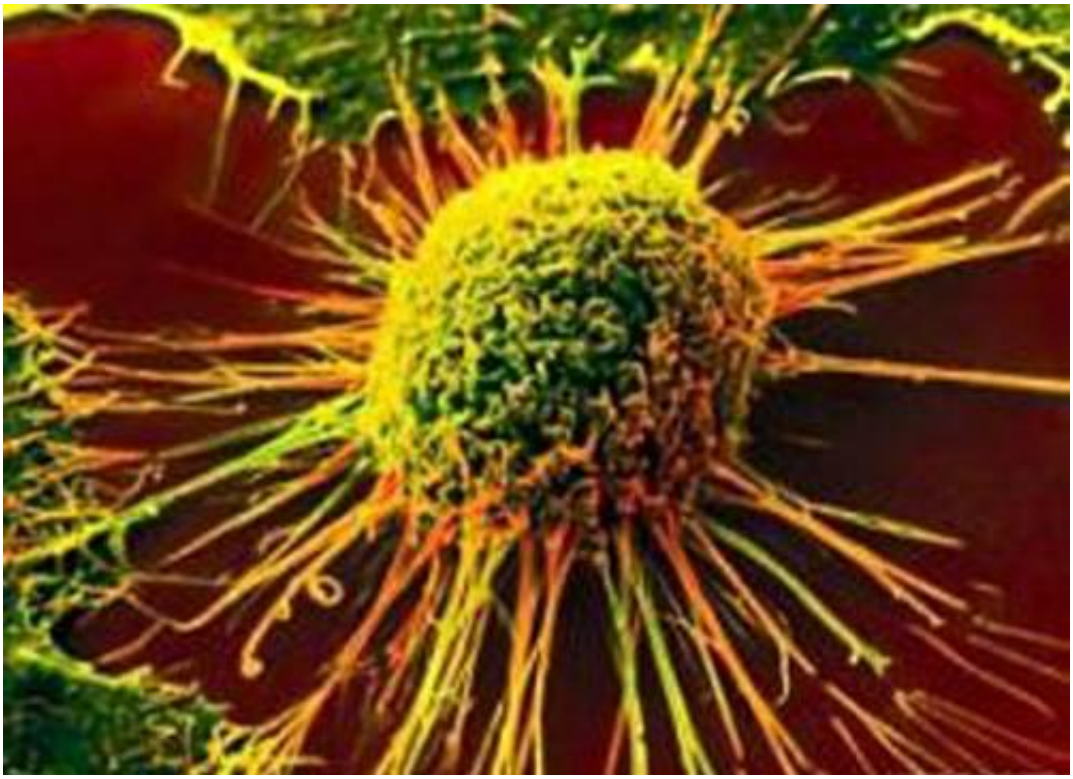

A non-invasive, low cost blood test that could help doctors diagnose some types of malignant childhood tumour has been developed by researchers at the University of Cambridge and Addenbrooke's Hospital, Cambridge University Health NHS Foundation Trust.

Reported today in the *British Journal of Cancer*, the test could enable doctors to monitor the effectiveness of treatments without exposing patients to repeated doses of radiation.

The target of the test is a type of cancer known as germ cell cancer. Germ cells are those cells in the body that go on to develop into sperm and egg cells. Germ cells can develop into tumours - both benign and malignant - particularly in the testes or ovaries, where the cells are normally found. However, occasionally germ cells can get trapped in the wrong part of the body during development and may later turn into brain tumours, for example.

The five year disease-free and overall survival rates for patients with high-risk malignant germ cell tumours remains less than 50%, and so accurate diagnosis and monitoring is crucial to improving outcomes for patients. All of the current tests are expensive, and none are ideal.

The most reliable diagnostic method currently in use is biopsy, where a section of the suspected tumour is extracted surgically and analysed by a pathologist. However, biopsies are prone to sampling errors and so may not be representative of the tumour as a whole. Computerised tomography (CT) scans and magnetic resonance imaging (MRI) also provide useful information but are not diagnostic and do not discriminate between benign and malignant tumours.

The ideal tool for diagnosis would be a non-invasive blood test; however, currently available tests only identify around three in five malignant germ cell tumours, potentially delaying diagnosis and the ability to prioritise patients for surgery. Accurate disease monitoring with routine blood testing is not possible for two in five patients, requiring follow up CT scans with exposure to harmful radiation and an associated increased secondary cancer risk.

"Although relatively rare, childhood germ cell tumours need to be diagnosed accurately and followed up carefully to give us the best chances of treating them," says Professor Nick Coleman from the Department of Pathology, University of Cambridge. "At the moment, we are not good enough at diagnosing these tumours and monitoring their treatment: we need better, safer and more cost-effective tests."

In research funded by Sparks charity, Great Ormond Street Hospital Children's Charity and Cancer Research UK, researchers at the University of Cambridge have developed a test for blood and cerebrospinal fluid samples that looks for a specific panel of four pieces of short genetic code known as microRNAs, which are found in greater quantities in malignant germ cell tumours. The test can distinguish malignant germ cell tumours from benign germ cell tumours and other cancers. The test can be used for diagnosis of malignant germ cell tumours in any part of the body, including the brain.

The test can also be used to check the effectiveness of treatments and, as it is safe and cost-effective, allows for frequent testing to monitor for the recurrence of malignant germ cell tumours.

Dr Matthew Murray from the Department of Paediatric Haematology and Oncology, Cambridge University Hospitals NHS Foundation Trust, says: "This test, developed with Dr Emma Bell, a postdoctoral scientist in our laboratory, could be exactly what we need: it could help us diagnose malignant germ cell tumours cheaply, safely and above all, more accurately than current methods. Our next step is to confirm our findings in a large clinical trial and, if this is successful, we hope to see the test in routine use in hospitals in the near future."

More information: Murray, MJ, Bell E, et al. A pipeline to quantify serum and cerebrospinal fluid microRNAs for diagnosis and detection of relapse in paediatric malignant germ-cell tumours. *British Journal of*

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