# New test for chronic blood cancers

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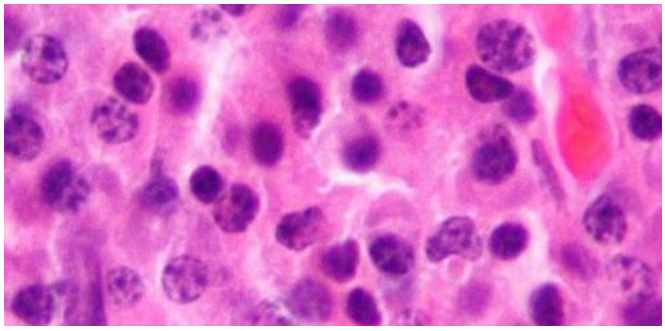

Micrograph of a plasmacytoma, a hematological malignancy. Credit: Nephron

(Medical Xpress)—A new test for blood cancers will catch many more cases than the present test that identifies only 60 per cent.

A simple blood test will soon be able to catch the vast majority of a group of chronic blood cancers, a new study reveals. Although around 60 per cent of cases can be identified with the current blood test, scientists did not know what caused the other cases and therefore could not test for it. Cambridge researchers have now identified a new cancer gene which accounts for the other 40 per cent of these chronic blood cancers. The research was published today, 10 December, in the *New England Journal of Medicine*.

Professor Tony Green, from the University of Cambridge's Cambridge Institute for Medical Research and Department of Haematology, who led the research said: "Diagnosing these chronic blood cancers is currently difficult and requires multiple tests, some of which are invasive and painful. Now, most patients with a suspected blood cancer will be able to be given a diagnosis after a simple blood test."

This group of chronic blood cancers – which affect an estimated 30,000 people annually in the UK – cause the over-production of red blood cells and platelets. These changes result in an increased incidence of blood clots which can be devastating when strokes or heart attacks occur. Although many patients can live for years with few or no symptoms, in some patients the disorders can become more aggressive with time and may even develop into acute leukaemia.

In 2005 scientists identified the JAK2 gene, mutationt in which are associated with around 60 per cent of blood cell disorders. Based on these findings a blood test was developed which transformed the way these blood disorders are diagnosed. Unfortunately, because the gene was only found in a little over half of people with chronic blood cancers, individuals who tested negative for the JAK2 gene would then have to undergo a battery of protracted, invasive testing to determine if they indeed had one of these disorders.

In the new study, led by the University of Cambridge and the Wellcome Trust Sanger Institute and supported by Leukaemia & Lymphoma Research together with the Kay Kendall Leukaemia Fund, scientists identified a new gene, CALR, which is altered in the other 40 per cent of blood disorders. For the research, the scientists sequenced the DNA of patients with chronic blood disorders. By analysing the DNA sequence, they were able to identify CALR as a new cancer gene which, when mutated, results in chronic blood cancers. Additionally, they found that patients with the CALR mutation – unlike those with the JAK2 mutation – had higher platelet counts and lower haemoglobin levels.

Peter Campbell from the Sanger Institute, who co-led the research, said: "There is now a sense of completeness with these disorders – the vast majority of our patients can now have a definitive genetic diagnosis made. In the next year or two, we will see these genetic technologies increasingly used in the diagnosis of all cancers, especially blood cancers."

Dr Jyoti Nangalia co-first author of the study from

the University of Cambridge said: "Not only will the identification of CALR lead to a new, less invasive test, we also hope that it can lead to new treatments – just as the discovery of JAK2 did. The CALR gene is involved in a cell function – aiding with the folding of proteins made by the cell - which has not implicated in these disorders before, so our research raises as many questions as it answers."

Provided by University of Cambridge

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