

Scientists uncover gene associated with blood cancers

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A genomic study of chronic blood cancer - a precursor to leukaemia - has discovered gene mutations that could enable diagnosis using only a blood test, avoiding the need for an invasive and painful bone marrow biopsy.

Researchers at the Wellcome Trust Sanger Institute identified the SF3B1 gene as being frequently mutated in myelodysplasia, one of the most common forms of [blood cancer](#). Myelodysplasia is particularly prevalent among people over the age of 60, and often the only symptom is anaemia, which makes it a challenge to give a positive diagnosis. Patients with [mutations](#) in the SF3B1 gene frequently had a specific abnormality of [red blood cells](#) in their [bone marrow](#), called ring sideroblasts.

The findings have significant potential for [clinical benefit](#), as the disease is often under-diagnosed. It is hoped that patients will soon be able to be screened for mutations in the SF3B1 gene through a single blood test.

"This discovery illustrates the promise of genome sequencing in cancer," says Dr Elli Papaemmanuil, lead author from the Sanger Institute. "We believe that by identifying SF3B1, and working to characterize the underlying biology of this disease, we will be able to build improved diagnosis and treatment protocols.

"Significantly, our analysis showed that patients with the SF3B1 mutation had a better overall chance of survival compared to those without the mutation. This suggests that the SF3B1 mutations drive a

more benign form of myelodysplasia."

In order to piece together the genomic architecture of myelodysplasia, the team sequenced all genes in the genome of nine patients with the disease. To their surprise, six had mutations in the SF3B1 gene. To expand their analysis, the researchers sequenced the SF3B1 gene in 2,087 samples across many common cancers.

In myelodysplasia, SF3B1 mutations were found in 20.3 per cent of all patients, and in 65 per cent of those patients with ring sideroblasts, making it one of the most frequently mutated [genes](#) so far discovered in this disease. Researchers found mutations of the same gene in up to 5 per cent of a range of other common cancers, such as other leukaemias, breast cancer and kidney cancer.

"Anaemia affects 1 in 10 people over the age of 65, and we cannot easily find a cause for the anaemia in a third of cases," says Peter Campbell, senior author, from the Sanger Institute and a practicing Haematologist at Addenbrooke's Hospital, Cambridge. "To diagnose myelodysplasia, we often have to resort to an invasive and painful bone marrow biopsy, but we hope this and future genetic insights will provide more straightforward diagnosis for patients through a simple [blood test](#)."

"Ever since I first saw these unusual and damaging [blood cells](#) - ring sideroblasts - down the microscope while training to become a haematologist, I have been fascinated by them and determined to find a cause. To discover a major genetic clue to their origins is very exciting, and I look forward to piecing together how the mutations cause these curious cells to develop and lead to this disease."

The SF3B1 gene encodes a core component of RNA splicing, an important editing mechanism that controls how the genome's message is delivered to the cell. The team discovered a strong association between

the gene and the presence of ring sideroblasts, making it the first gene to be strongly associated with a specific feature of the disease. Ring sideroblasts are abnormal precursors to mature red blood cells with a partial or complete ring of iron-laden mitochondria (energy generators) surrounding the nucleus of the cell. Their presence is frequently associated with anaemia.

"These genetic discoveries are very important and could potentially assist clinicians when diagnosing blood cancers in patients, avoiding the need for invasive bone marrow biopsies. Myelodysplasia is becoming increasingly prevalent in people over 60, and cases will continue to rise with our increasing ageing population, particularly among those who suffer from [anaemia](#)," says Dr David Grant, Scientific Director at [Leukaemia](#) & Lymphoma Research. "We are delighted to have supported research into the genomic architecture of myelodysplasia which will contribute towards making a difference for the diagnosis and treatment for patients."

Provided by Wellcome Trust Sanger Institute

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