Antibody genes influence forgotten heart disease

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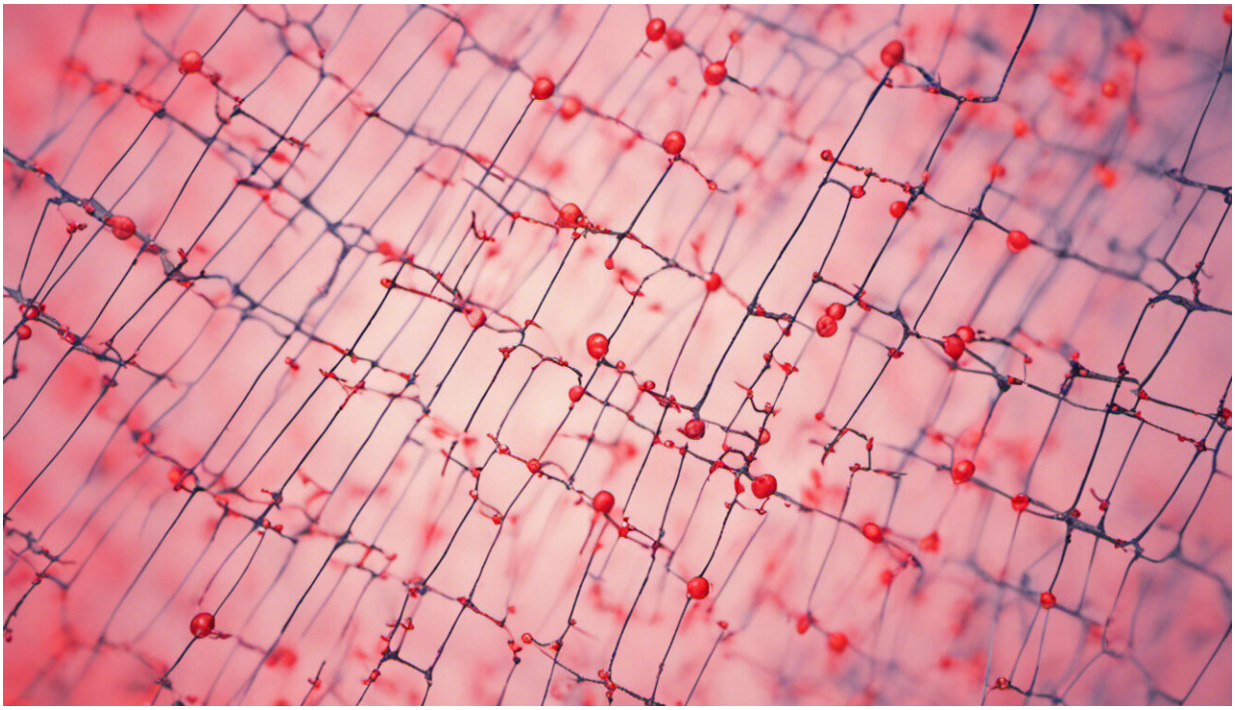

Credit: AI-generated image (disclaimer)

New research has found that genetic differences in antibody genes alter individuals' susceptibility to rheumatic heart disease, a forgotten inflammatory heart condition – known as 'RHD' – that is rife in developing countries.

Rheumatic heart disease is triggered by bacterial sore throat or skin infection during childhood. The disease causes the heart valves to stiffen until eventually the heart starts to fail. Without surgery, which is usually not available to those affected, many will die during early adulthood.

The disease is endemic in the Pacific and in Fiji, where the disease burden has been most comprehensively studied, it is a leading cause of death in the young causing a similar number of deaths among adolescents and young adults to suicide and road traffic accidents. Several studies have shown the disease is also found frequently in Sub-Saharan Africa. While estimating the death toll there has been more difficult, it is likely to be substantial.

Remarkably, not long ago, rheumatic heart disease was a major public health concern in the UK, Europe and USA. But as living conditions improved, the disease disappeared and it was all but forgotten by doctors and the public. Crucially, as a consequence, little research has been done for decades on why people get the disease. Moreover, prevention is currently very difficult and there is no vaccine.

Now doctors and scientists from Oxford University, working in collaboration with researchers in the Pacific, France, USA and Australia, have taken an important step to defining why some people get the disease while others do not. Their work involved samples from over 3,000 individuals recruited across the Pacific with the largest number of samples coming from Fiji and New Caledonia. The team then did genetic analysis including sequencing and analysed the data using super computers at the University of Oxford.

The research found that the code for a certain antibody gene differed in those with and without the disease. This finding is surprising and important because in recent years antibody genes have received little attention from those who study inflammatory or autoimmune disease, so

it may have ramifications beyond rheumatic heart disease.

Prof Adrian Hill at the University of Oxford, one of the project's leaders, said: 'This study provides new insights into the role of antibody genes in rheumatic heart disease. This could provide important clues for development of an effective vaccine against the bacteria that trigger this very neglected disease.'

Associate Prof Andrew Steer at the Murdoch Children's Research Institute, another of the project's leaders, said: 'The burden of death and disability associated with rheumatic heart disease in developing countries worldwide is astonishing. So it's important we do high quality research to better understand this disease going forward.'

Also Dr Joseph Kado, a paediatric cardiologist from Fiji National University, who lead the study in Fiji, said: 'In Fiji it is not uncommon to see children and young adults die from rheumatic heart disease. It is therefore important that Fiji participates in international efforts to tackle the disease including the latest research techniques in human genetics.'

More information: Tom Parks et al. Association between a common immunoglobulin heavy chain allele and rheumatic heart disease risk in Oceania, *Nature Communications* (2017). DOI: 10.1038/ncomms14946

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