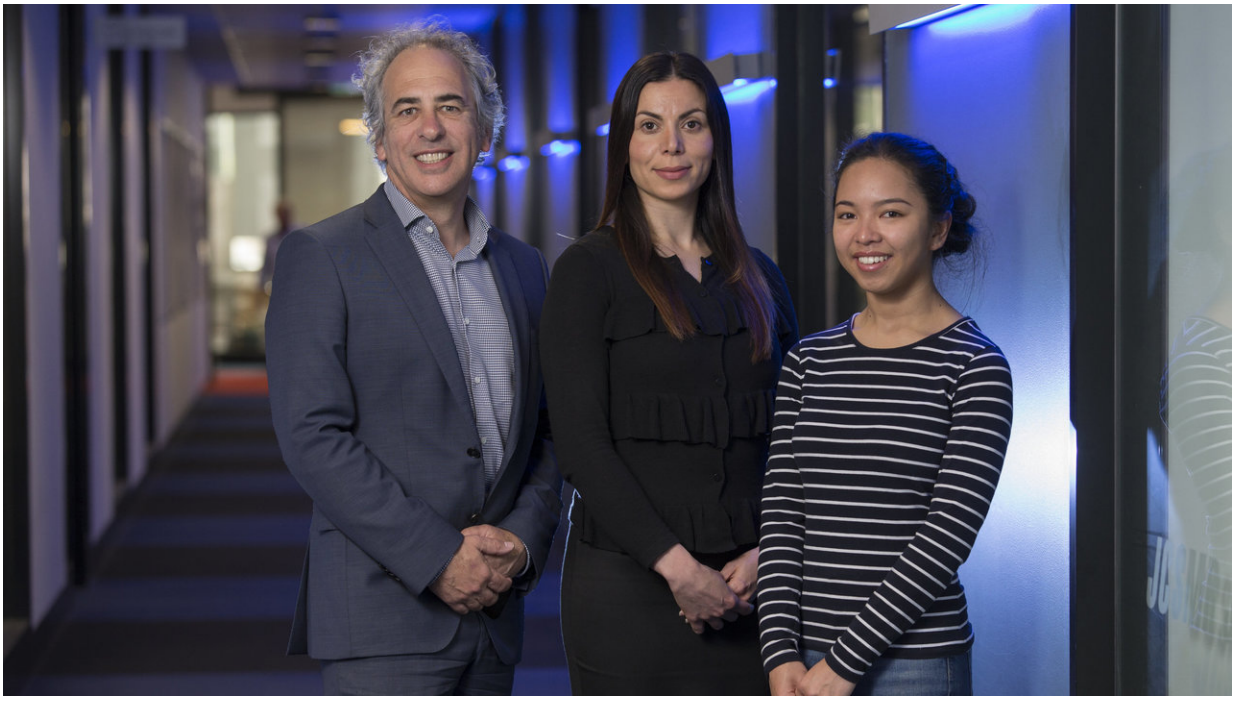
Researchers find new genetic disease

October 18 2018



Credit: Australian National University

Researchers at The Australian National University (ANU) have discovered a new genetic disease and a method for detecting more unexplained medical conditions.

"We've discovered a new syndrome, the genetic cause and the mechanism that explains how the genetic variation causes the illness," said Professor Matthew Cook from The ANU College of Health and

Medicine.

"It is a disease which is characterised by immune deficiency where patients get recurrent infections, especially chest infections, including recurrent pneumonia, but paradoxically patients also suffer inflammatory diseases of the skin, lymph nodes and the spleen," said Professor Cook.

"Some patients with the condition have been unwell for some 20 years and until now have not had a definite diagnosis."

Over the past five years, a dedicated team of researchers at the Centre for Personalised Immunology in the ANU College of Health and Medicine has been gene-hunting to find insights into rare diseases.

"Human genome sequencing is now relatively straightforward but the big challenge is to understand how genetic changes cause disease," said Professor Cook.

"We study rare diseases of the immune system. The methods employed in this study can be applied to resolve otherwise unexplained diseases."

The symptoms affecting patients with this unnamed syndrome are a combination of the immune system being too weak and not mounting a proper response, while in other respects producing an excessive response.

The research team identified the condition by whole genome sequencing patients and then characterising the patients' immune systems.

Meanwhile research colleagues in Japan identified the same disease in a second family with the same genetic variant.

"The Japanese patients had similar clinical problems and it turned out they had exactly the same genetic variant," said Professor Cook.

"To be sure we were dealing with the correct gene, we went on to introduce exactly the same genetic change in a mouse model using gene editing. This combined approach provides a powerful method for resolving potentially important genetic variants."

The discovery offers hope to other patients suffering from rare conditions who have struggled with getting a definitive diagnosis and appropriate treatment.

"Solving rare diseases is very important for two reasons. First, a genetic diagnosis provides certainty to patients who may have previously undergone many tests over many years, often without obtaining an answer," Professor Cook said.

"Second, rare diseases can provide important information about how the body works that can be useful for developing new tests and treatments for other diseases.

"Patients can now get some certainty which makes a big difference to people and we can use the process again and again."

Provided by Australian National University