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Discovery may ease the agony of lupus

By **NATASHA ROBINSON**, HEALTH EDITOR
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Researchers have for the first time identified a gene which when overactivated is responsible for causing the auto-immune disease lupus.

The gene TLR7 is programmed to help the immune system guard against viral infections, but in its mutated form it can become aggressive and cause the immune system to attack healthy cells.

The discovery has been made by Australian National University and Hudson Institute of Medical Research scientists, and the findings are being published in the prestigious journal Nature.

About 20,000 people in Australia have lupus, which affects nine times as many women as men. The disease is characterised by inflammation which can affect the skin, joints, and less commonly the blood and organs including the lungs, kidney and heart. Sufferers commonly experience rashes, joint pain and extreme tiredness. There is currently no cure for lupus.

Researchers studied a group of patients with lupus and identified a mutation of the TLR7 gene in a Spanish girl, Gabriella, who was diagnosed with severe lupus when she was seven years old.

Such a severe case with early onset of symptoms is rare and indicates a single genetic cause.

In a laboratory at the Centre for Personalised Immunology at ANU, researchers identified a mutation of the TLR7 gene in Gabriella's case, and identified other cases of severe lupus in the group of patients studied where the TLR7 gene was also mutated.

They then induced the TLR7 gene mutation in mice, and the mice developed lupus.

TLR7 is a critical sensor of our immune system. It is set off by viral and bacterial RNA produced during an infection.

Once engaged, TLR7 activates a suite of responses that facilitate clearance of the pathogen by immune cells – an inflammatory reaction that is normally transient.

The discovery of the gene's role in lupus has implications for the future treatment of lupus and will pave the way for development of new drugs to treat the disease, says study author Vicki Athanasopoulos from the John Curtin School of Medical Research at ANU.

Current drugs are predominantly immune suppressors and can cause significant side effects.

"We now have a platform that we can use to try and develop more effective and targeted treatments to try and treat lupus and also other auto-immune diseases, because we suspect that this gene may also play a role in driving other auto-immune disease," Dr Athanasopoulos said.

"The next steps is to try and repurpose current medications, and trialling some novel drugs."

NATASHA ROBINSON, HEALTH EDITOR

Natasha Robinson began her career at The Australian in 2004. A Walkley awards finalist and a Kennedy Awards winner, she was appointed Health Editor in 2019, and has covered rounds including national affairs, in... Read more