

# Investigating genetic links to common autoimmune diseases

Autoimmune diseases such as rheumatoid arthritis and Graves' disease affect around five per cent of the population and can be both debilitating and fatal.

Autoimmunity is partly caused by inheritance of a combination of particular gene variants. A research team based at the University of Otago has previously played a role in the identification of one such variant (PTPN22).

In the hunt for more variants, an international collaboration has been formed to combine data and validate results with other research institutions.

The New Zealand arm of the study is funded by the HRC and led by Dr Tony Merriman at the University of Otago and international partners include Dr Simon Pearce from the University of Newcastle-upon-Tyne, Dr Sophia Steer from King's College London and Dr Timothy Vyse from Hammersmith Hospital in London.

Each research team has an independent cohort which they have been following to identify gene variants associated with autoimmune diseases. This has involved testing every gene in the human genome, using SNPs (single nucleotide polymorphism) as surrogate markers. Data from these cohorts will be combined and common gene variants will be further tested for validation.

"We have all the information, but we need to combine it and see if any genes come to the top of the list," says Dr Merriman.

Dr Merriman says information gathered from individual cohorts can produce false-positives, where an association of a certain gene with an autoimmune disorder has occurred by chance. Combining the cohorts and then validating the results in independent cohorts will allow researchers to focus on testing genes that are more likely to hold the answers.

The research team will look at a higher density of SNP markers within genes they validate in order to identify the precise disease-causing genetic variant.

Dr Merriman says the international collaboration also provides the New Zealand team with access to genotyping technology not available in New Zealand. The team will be able to use the Sequenom MassARRAY genotyping machine at the University of Newcastle-upon-Tyne. This represents a huge advancement to the technology currently available in New Zealand and will allow the team to analyse hundreds of genetic variants at once.

The research has the potential to improve treatments for autoimmune disease by helping to develop more targeted therapies for the one in 50 New Zealand adults affected by autoimmune disease.

This research has been funded through Objective One of the International Investment Opportunities Fund. The fund enables outstanding New Zealand research teams to build research collaborations with overseas research teams and participate in high-end research which will benefit the health of New Zealanders.

Through this project Dr Merriman and his team hope to share technology and ideas and raise New Zealand team's profile. In turn, there is potential for further external funding in the future.

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## **Key words:**

Autoimmune disease, gene variants

## **Key facts:**

- Autoimmune disease affects one in 50 New Zealanders
- Common autoimmune diseases include: rheumatoid arthritis, Graves' disease and systemic lupus erythematosus and diabetes mellitus type 1
- Autoimmunity is partly caused by inheritance of certain gene variants
- Environment is also important in determining risk of autoimmune diseases, but knowledge on the exact environmental factors is scarce. However, it is known that smoking can increase the risk of rheumatoid arthritis.

## **Aims of this research:**

- To validate current findings from the New Zealand cohort study with other international cohorts
- To build international partnerships with UK research teams in order to share information on autoimmune disorders
- To raise the profile of the research undertaken in New Zealand.