

## New Australian study investigates the effect of genetic change on immune cells

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Dr Judith Field

There are now over 110 genetic changes that are known to increase an individual's susceptibility to MS. Current genetic research has now moved on to investigating each of these genetic changes in more detail to try and determine the mechanisms that underlie the MS disease process with a view to identifying new therapeutic pathways.

A new Australian study, led by Dr Judith Field from the University of Melbourne and coordinated through the [ANZgene](#) group has recently been published in the medical journal *PLoS One*.

ANZgene is one of MS Research Australia's research platforms which concentrates on the role of genetics in the development and disease course of MS. This collaborative group of researchers from Australia and New Zealand work on a number of genetic studies concurrently and also coordinate samples of genetic material and genetic data from people with MS for international collaboration.

The [new study](#) focuses on the effects of genetic change in a molecule known as CD40. The CD40 gene has previously been identified as a risk gene for MS as well as a number of other immune related diseases and the molecule functions in a number of immune cell types.

Dr Field and her team showed that the genetic change that increases the risk of developing MS leads to lower levels of CD40 in immune cell subtypes known as B cells and dendritic cells. When the risk gene was present the ratio of CD40 on the cell surface compared with the secreted form of CD40 was also different in immune cells. This work demonstrates the way that genetic changes in the CD40 gene can effect immune cell function and may impact on the development of MS in individuals.

The research team also showed that irrespective of the form of the CD40 gene, people with MS had lower levels of CD40 in B cells compared with healthy controls. CD40 may be playing a role in the immune regulation that protects against MS and this function is impaired by this genetic change.

These types of studies are important to uncover the mechanisms of MS and through this find new pathways for treatment.