

## Australian study identifies distinct ‘methylation signature’ in immune cells

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It is known that people develop MS due to a combination of risk genes and environmental factors. One way that environmental factors can interact with genes is through methylation. Methylation molecules tag genetic sequences and control their activity levels – whether genes are switched on or off in certain tissues or in response to certain circumstances in the environment. Both smoking and vitamin D are known to affect methylation patterns.

Since MS results from the interface between genes and the environment, looking at the methylation patterns of genes in people with MS is likely to reveal clues about what causes or propagates the disease.

A group of Australian researchers, led by [Dr Vicki Maltby](#) from the Hunter Medical Research Institute and funded by MS Research Australia, have investigated methylation patterns in a subset of immune cells thought to be involved in MS. These cells, known as CD8+ T cells, were taken from people with relapsing-remitting MS and compared to cells taken from healthy individuals.

Using an experimental technique that allowed the researchers to look at the methylation patterns at all genes at once, they found that the immune cells in people with MS had a specific ‘signature’ that differentiated them from healthy cells. Published in the scientific journal [Clinical Epigenetics](#), the methylation patterns seen in CD8+ T cells were different to those seen in other immune cell subtypes.

[Previous work](#) in this laboratory investigated the methylation signals in another immune cell subset that is thought to be involved in MS. This earlier study looked at CD4+ T cells, which have long been thought to be a driving cell behind the disease process in MS. The researchers identified a distinct methylation difference at the major MS risk gene in people with MS compared with healthy controls. This gene is called HLA-DRB1\*15 and is responsible for most of the genetic risk of developing MS.

Both studies used the same patient and healthy control group allowing for a direct comparison of the findings. Analyses such as this are complicated but can be very valuable in determining the role of the different factors in MS. The complex statistical analysis undertaken in this study, by Dr Rod Lea, was also funded through the MS Research Australia Bioinformatics Fellowship. [Read more about Dr Lea’s work.](#)



Methylation patterns provide a link between environmental exposures in a person's lifetime and the activities of their genes. Research in this area will provide further insights into the way factors in the environment act on genes to cause or exacerbate MS. This study forms part of a wider research interest in this area at the Hunter Medical Research Institute led by Associate Professor Jeannette Lechner-Scott. More information about the research being carried out by this group can be found [here](#) and [here](#).