Two new studies identify genetic changes linked to specific symptoms of MS

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Over the last few years, genetic studies that included thousands of people with MS from around the world have identified a large number of genetic changes that may increase a person's risk of developing MS (see our previous reports <u>here</u> and <u>here</u>). Over 110 genes are now known to be part of the MS story.

Now that these genetic changes are identified, ongoing studies are concentrating on investigating the role of these changes in more detail. Two recent studies have done just that, to try and uncover clues that will enable the use genetics to better predict the clinical course of someone's MS or discriminate between treatment choices.

The <u>first study</u> published in the medical journal *PLoS One*, looked at the 57 genetic variations that were identified in 2011. They confirmed a number of the genetic findings of the <u>original study</u> and also found that combinations of genetic changes were associated with a higher risk of developing MS than the single changes alone. This finding seems to confirm the current thinking that multiple genetic changes in an individual add together to determine a person's susceptibility to MS.

This study also showed that particular symptoms of MS were associated with combinations of genetic change. Specifically, they showed visual impairments due to optic neuritis, difficulties with voluntary muscle coordination (such as walking) and muscle weakness were linked to different genetic changes.

In the <u>second study</u> published in the journal *Multiple Sclerosis*, researchers calculated the cumulative genetic risk score using the 110 genetic changes known to increase MS risk for 842 people with MS and compared this with 321 healthy people. As expected, people with MS had a higher genetic risk than people without the disease. When specifics about people's MS were examined, people who had positive result for oligoclonal bands in their cerebrospinal fluid (a diagnostic test for MS that checks for proteins related to inflammation in the cerebrospinal fluid) had a higher genetic risk than those for whom the test was negative. This was true particularly for genetic risk at the HLA (Human Leukocyte Antigen) region, which is the main risk gene for MS. They also found that the IgG index, another measure of inflammatory proteins, was associated with HLA genetic risk.

Contrastingly, they found that relapse rate was associated with the non-HLA component of genetic risk. People with higher non-HLA genetic risk had both an increased relapse rate and a shorter period without a relapse after the onset of their disease.

If these findings are confirmed, this type genetic information could be used in future to predict symptoms in individuals with MS and tailor treatment.