

Genetic variant tied to MS and systemic lupus identified

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(HealthDay)—A genetic variant that is associated with both multiple

sclerosis and systemic lupus erythematosus (SLE) has been identified, according to a study published online April 26 in the *New England Journal of Medicine*.

Maristella Steri, Ph.D., from Istituto di Ricerca Genetica e Biomedica in Italy, and colleagues conducted a genome-wide association study in [multiple sclerosis](#) as well as *TNFSF13B* locus-specific association testing in SLE. To identify the causal variant and determine its mechanism of action, they performed extensive phenotyping of quantitative immune variables, sequence-based fine mapping, cross-population and cross-phenotype analyses, and gene expression studies.

The researchers found that a variant in *TNFSF13B*, encoding the cytokine and drug target B-cell activating factor (BAFF), was associated with both multiple sclerosis and SLE. There was also an association between the disease-risk allele and up-regulated humoral immunity through increased levels of soluble BAFF, B lymphocytes, and immunoglobulins. An insertion-deletion variant, GCTGT→A (in which A is the risk allele) was identified as the causal variant and yielded a shorter transcript that escaped microRNA inhibition and increased production of soluble BAFF. This in turn up-regulated humoral immunity. This autoimmunity [variant](#) has been evolutionarily advantageous, based on population genetic signatures, most likely by augmenting resistance to malaria.

"Overall, the evolutionary scenario we propose is that BAFF-var was selected as an adaptive response to malaria infection, resulting in an increased present-day risk of autoimmunity," conclude the authors.

More information: [Abstract/Full Text \(subscription or payment may be required\)](#)

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