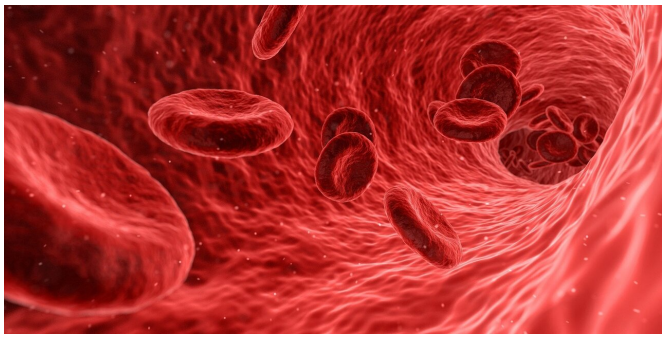
Immune cell-related genetic variants that may impact autoimmune conditions isolated

15 September 2020, by Bob Yirka



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A team of researchers from Italy, the U.K. and the U.S. has isolated a number of immune cell-related genetic variants that may impact autoimmune conditions. In their paper published in the journal *Nature Genetics*, the group describes the host of tools and techniques they used to uncover previously unknown immune-cell variants that might be associated with a variety of immune-related diseases.

Prior research has shown that a large number of diseases once thought to be organ- or site-specific also have immune response connections. Inflammatory bowel disease, multiple sclerosis and Kawasaki disease are just a few examples. In response to such findings, researchers have been taking a harder look at immune cells and associated variants that might be behind the onset of such diseases.

The work involved conducting genome-wide association studies (GWAS) on blood samples from 3,500 people living in Sardinia. The studies involved the use of genotyping profiles and imputation as a means of looking for variants involving 731 immune cell traits. The actual work involved using cell surface markers and flow cytometry-based cell sorting.

In all, the researchers looked at data for 22 million variants found in the blood samples (some of which came from people in the same family). In so doing they found 122 single nucleotide polymorphisms, 52 of which were new and 17 loci that could be independently tied to 459 individual cell traits. They note that the set also included variants found at approximately 36 loci that had been associated with known autoimmune diseases.

The researchers also conducted fine-mapping, follow-up validation, selection analyses, expression and quantitative trait locus to gain a better perspective on the association traits they had isolated—and more importantly, to see if they could find any previously unknown associations to autoimmune diseases. They next plan to investigate the possibility of using protein quantitative trait loci that they identified to find any connections between their findings and drugs already in use—and also to see if such drugs might be used for other conditions. They note that going forward, it might a good idea to target more than one pathway or protein during therapy development based on immune cell subtypes and the diseases involved.

More information: Valeria Orrù et al. Complex genetic signatures in immune cells underlie autoimmunity and inform therapy, *Nature Genetics* (2020). DOI: 10.1038/s41588-020-0684-4