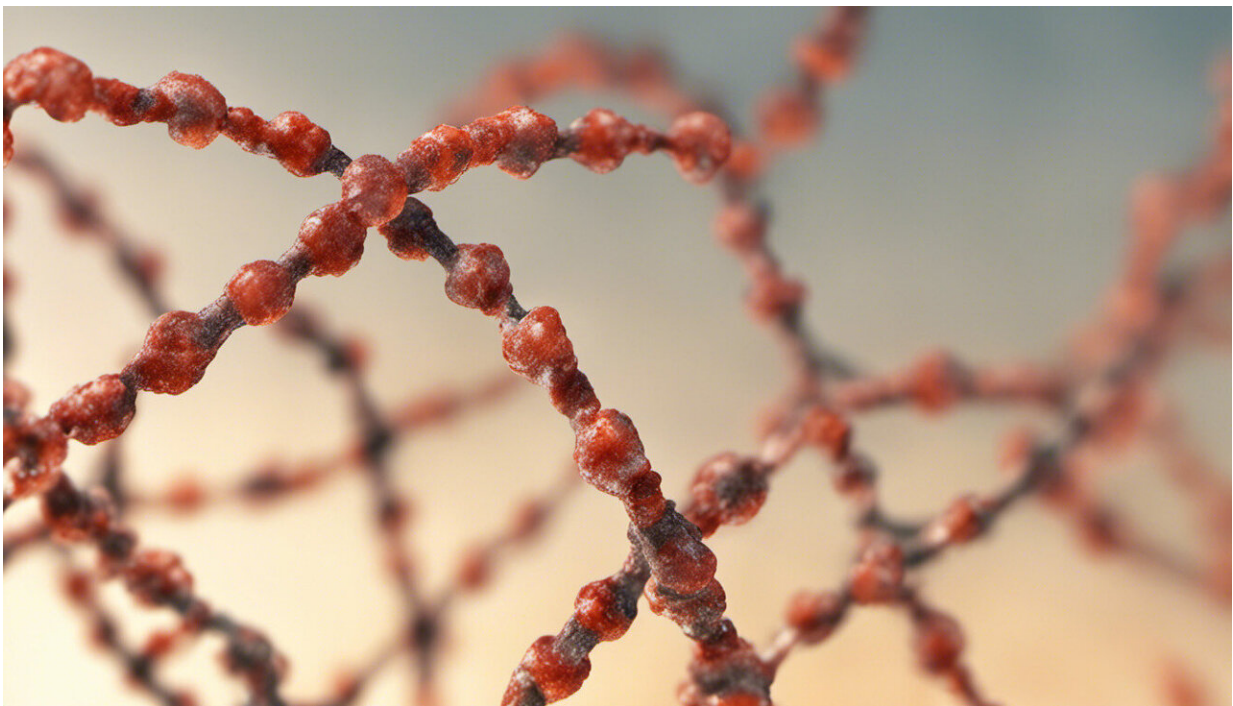


Genome-wide association study identifies six new susceptibility loci tied to the age-old disease of leprosy

May 27 2015



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A*STAR researchers have conducted a genome-wide association study that has important implications for the early diagnosis, and therefore prevention, of leprosy—one of the oldest documented diseases in human history.

Leprosy is a chronic bacterial infection caused by the bacterium *Mycobacterium leprae* that primarily targets the skin and peripheral nerves. The ailment typically results in skin lesions, immune impairment and sensory loss that, if left untreated, can lead to permanent skin damage, severe complications, as well as physical deformities. In fact, leprosy patients have been known to lose their fingers and toes because they lack the ability to sense environmental dangers and thereby avoid physical injuries.

The prevalence of leprosy has declined dramatically thanks to the implementation of multidrug therapy by the World Health Organization in the 1980s. But the disease remains a major public health problem, given that more than 200,000 new cases are still being detected worldwide annually.

Jianjun Liu and co-workers at the A*STAR Genome Institute of Singapore, together with researchers in China and the United States, have now conducted a genome-wide association study in the Chinese population and identified a great number of susceptibility loci—or clusters of genes—that render an individual more vulnerable to the disease.

Liu and colleagues analyzed and compared the genetic profiles of 24,330 individuals from various regions of China, of which 16,017 were controls (healthy individuals) and 8,313 were cases (individuals with leprosy). They identified 16 susceptibility loci that were frequently present in cases but not in controls. Included among these were six novel susceptibility loci, discovered for the first time in the Chinese population.

The researchers investigated the biological and functional relevance of the genes within the confirmed loci. They identified BATF3, CCDC88B and CIITA, which are involved in immunity, as candidate susceptibility

genes for leprosy. Furthermore, they carried out a systematic evaluation of the pleiotropic effects of the susceptibility loci by searching for reported associations with other diseases. They found that of the 16 established [leprosy](#) loci, 11 had reported associations with autoimmune and inflammatory diseases, such as sarcoidosis and [inflammatory bowel disease](#).

"By discovering a good number of genes that can affect the risk of getting the disease, we can develop better strategies for [early diagnosis](#) and prevention, which is essential for permanently eradicating the disease," says Liu.

More information: Discovery of six new susceptibility loci and analysis of pleiotropic effects in leprosy. *Nature Genetics* 47, 267–271 (2015). [dx.doi.org/10.1038/ng.3212](https://doi.org/10.1038/ng.3212)

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