Mystery of immune system infections slowly unravels

The mutations were familiar, but the patients' conditions seemed baffling at first. A team lead by Rockefeller University researchers had linked variations in an immune gene to rare bacterial infections.

Shortly afterward, Chinese scientists told them of three children in that country with mutated versions of the same gene. However, the Chinese children had no history of the severe bacterial infections. Instead, they had seizures and unusual calcium deposits deep in their brains.

This discrepancy led to the discovery of an immune protein with paradoxical roles: It both aids and tamps down aspects of an immune system response.

"It has turned out that mutations in a single gene eliminate the immune protein ISG15, giving rise to two different problems: an inability to resolve harmful inflammation, which can lead to autoimmune disease, and susceptibility to infections caused by the tuberculosis bacterium and its cousins," Casanova says. "By identifying the source of this genetic disorder, we have taken a first step toward finding treatments for those facing the autoimmune disease and severe TB-related infections it may produce."

Read full, original article: Single gene links susceptibility to rare infections with predisposition to autoimmune disease