

Development of autoimmunity in patients with common variable immune deficiency

September 24 2013

Common variable immune deficiency (CVID) is a genetic disease associated with enhanced susceptibility to infection, autoimmunity, and decreased antibody production. Mutations in the tumor necrosis factor receptor superfamily member *TACI*, are associated with CVID and autoimmunity development. Interestingly, autoimmunity develops in CVID patients with only one mutated copy of *TACI*, and CVID patients with two mutated *TACI* alleles do not develop autoimmunity.

In this issue of the *Journal of Clinical Investigation*, Eric Meffre and colleagues at Yale University evaluated B cell activation and tolerance development in healthy individuals and CVID patients with one or two mutated copies of *TACI*. The authors found that CVID patients with a single altered *TACI* allele maintained some residual B cell responsiveness that promoted development of autoantibodies, whereas individuals with 2 mutated copies of *TACI* have complete impairment of B cell responses, which likely prevents autoimmunity.

In the companion commentary, Antonia La Cava of the University of California Los Angeles suggests that targeting residual B cell activity in CVID patients that are heterozygous for *TACI* mutations may provide clinical relief.

More information: CVID-associated TACI mutations affect autoreactive B cell selection and activation, *J Clin Invest.* [DOI: 10.1172/JCI69854](https://doi.org/10.1172/JCI69854)

Provided by Journal of Clinical Investigation

Citation: Development of autoimmunity in patients with common variable immune deficiency (2013, September 24) retrieved 2 May 2024 from <https://medicalxpress.com/news/2013-09-autoimmunity-patients-common-variable-immune.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.