Introduction

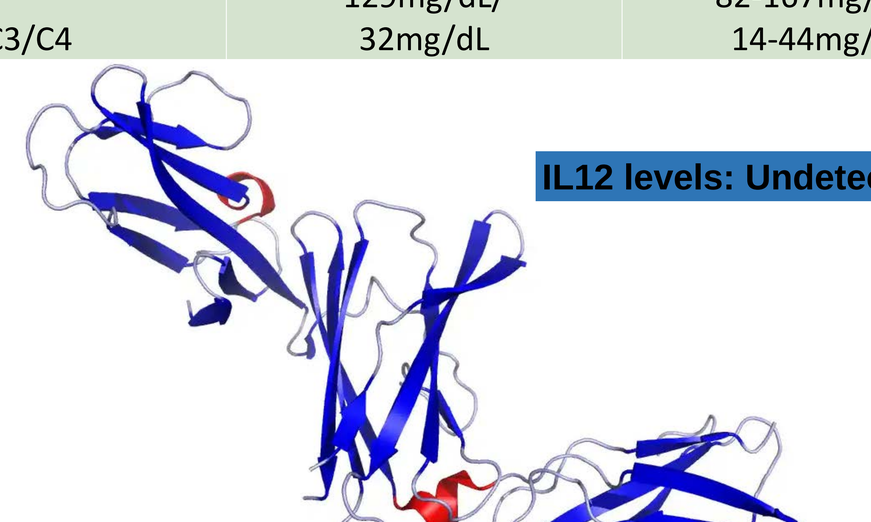
Mendelian Susceptibility to Mycobacterial Disease (MSMD) is a diverse group of diagnoses related to impairment in the immune response to intracellular pathogens, which can be mediated by defects at many points along the way in the mononuclear phagocyte/T helper cell type 1 axis. Symptoms typically present in childhood with infection caused by non-pathogenic mycobacteria such as the BCG vaccine. Immunodeficiency 29 is the subset of MSMD caused by mutations in IL-12. We describe a novel mutation causing Immunodeficiency 29 in an Active Duty patient.

Case Report

A 38y/o female was evaluated for frequent diarrheal illnesses and upper respiratory infections with prolonged treatment courses. Initial evaluation was concerning for common variable immune deficiency based on low immunoglobulin levels. Response to protein vaccines was normal, while response to polysaccharide vaccines was impaired. History concerning for recurrent infection with intracellular pathogens, including *Salmonella,* was not classic for CVID, so further evaluation with genetic testing was undertaken. This revealed a 5 base-pair duplication causing a frameshift mutation in the *IL12B* gene (p.TyrArgfs*59). Further testing revealed undetectable levels of IL12, consistent with Immunodeficiency 29.

*The opinions or assertions herein are the private views of the authors and are not to be construed as reflecting the views of the Department of the Air Force or the Department of Defense.

Lab Test	Result	Normal
IgA	212mg/dL	70-400mg/dL
IgG	**679mg/dL**	700-1600mg/dL
IgM	**29.4mg/dL**	40-230mg/dL
PCV Response	**43.50%**	>70%
Diphtheria Response	Positive	Positive
Tetanus Response	Positive	Positive
C3/C4	129mg/dL/ 32mg/dL	82-167mg/dL/ 14-44mg/dL



IL-12B Protein Crystal Structure[1]

IL12 levels: Undetectable

Case reports
of Middle E
loss-of-funct
typically resu
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generally he
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