Question » Does autoimmune cytopenia lead to an immune deficiency or vice versa?

I have autoimmune cytopenia and an immune deficiency, and I want to understand the relationship between them. I understand what each is, but does one lead to the other?

Abbie » I spoke with Terry O. Harville, MD, PhD, medical director of the Special Immunology Laboratory at the University of Arkansas for Medical Sciences, who explained there are several issues at work. First, in certain circumstances, the failing immune system retains parts of immunity that would normally be eliminated, since these could cause autoimmunity such as cytopenias. He compared this process to "letting prisoners out of jail to act as soldiers! It probably wouldn't work well." The failing immunity may be trying to do the best it can to fight infections, even at risk for autoimmunity.

Second, we now know there are approximately 400 gene mutations that can cause immunodeficiency, autoimmunity or both in the same person. Therefore, in some circumstances, cytopenias appear first, then the immunodeficiency or vice versa. Or, both may be discovered simultaneously. So, having cytopenias in addition to an immunodeficiency is not considered rare.

Following is a list of some of the gene mutations that can cause autoimmunity, immunodeficiency or both. Note there are several listed that can cause hematologic issues (cytopenias):

Gene Affected	Primary Immunodeficiency	Autoimmunity
AIRE	APECED	Polyendocrinopathy
BAFF-R	Common Variable Immunodeficiency	Hematologic, Other
BCL10	Antibody Deficiency	Gastrointestinal Disease
CD19	Common Variable Immunodeficiency	Hematologic, Glomerulonephritis, Other
CD81	Antibody Deficiency	Glomerulonephritis, Other
CTLA4	Antibody Deficiency	Lymphoproliferation, Other
FOXP3	IPEX	Immunodysregulation, Polyendocrinopathy and Enteropathy, X-linked
ICOS	Common Variable Immunodeficiency	Hematologic, Other
IL21	Antibody Deficiency	Colitis, Other
LRBA	Antibody Deficiency (IgG and IgA)	Inflammatory Bowel Disease
MSH-5	Common Variable Immunodeficiency	Hematologic, Other
NFKB2	Antibody Deficiency	Alopecia, Endocrine Adrenal
PIK3CD (p110)	Antibody Deficiency	Lymphoproliferation, Inflammatory Bowel Disease
TNFRSF5	Decreased IgG, IgA (IgM normal or increased)	Gastrointestinal Disease
TNFRSF7	Antibody Deficiency	Aplastic Anemia
TNFSF12	Decreased IgA and IgM	Glomerulonephritis, Other

For more information, go to primaryimmune.org/about-primary-immunodeficiencies/relevant-info/autoimmunity.

» *Have a question? Email us at editor@IGLiving.com. Your information will remain confidential unless permission is given.*

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