Research aids discovery of genetic immune disorder

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Investigators at the National Institutes of Health (NIH) and international colleagues have identified a genetic immune disorder characterized by increased susceptibility and poor immune control of Epstein-Barr virus (EBV) and, in some cases, an EBV-associated cancer called Hodgkin's lymphoma. The researchers studied two unrelated sets of siblings with similar immune problems and determined their symptoms were likely caused by a lack of CD70, a protein found on the surface of several types of immune cells. Scientists at the National Institute of Allergy and Infectious Diseases (NIAID), part of NIH, conducted the research with an international team of collaborators.

Both sets of siblings had evidence of uncontrolled infection with EBV, a common and usually mild virus, which resulted in the development of Hodgkin's lymphoma in three of these children. Each child also had other immune symptoms, such as reduced activity of pathogen-fighting T cells, low production of antibodies and poor activation of antibody-producing B cells.

The researchers analyzed the genomes of all four children and found that each had two mutated copies of the *CD70* gene, resulting in nonfunctioning or nonexistent CD70 proteins. All four parents, who had healthy immune systems, had only one copy of the mutation—indicating that CD70 deficiency follows an autosomal recessive pattern of inheritance. This means affected individuals receive a flawed gene from each parent in order to have symptoms. While no specific treatment for CD70 deficiency currently exists, each of the four children has

recovered from Hodgkin's lymphoma and is receiving antibody infusions to help bolster the immune system.

This work also offers insight into the normal role of CD70. Previous studies showed that CD70 interacts with another immune cell protein called CD27, an interaction that may be important for the proper function of lymphocytes. This hypothesis is affirmed by these latest findings on CD70 deficiency. The research also indicated that investigators testing experimental medications that decrease the activity of CD70 or CD27—a possible strategy for combatting autoimmune disease—should be aware of a possible risk of EBV-related complications.

More information: H Abolhassani et al. Combined Immunodeficiency and Epstein-Barr Virus-Induced B Cell Malignancy in Humans with Inherited CD70 Deficiency. *Journal of Experimental Medicine* DOI: 10.1084/jem.20160849 (2016).

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