Unlocking the Secrets of Autoimmunity

Mar 5, 2010

It is hard to believe that a single patient can lead to new insights into autoimmune disease that merit publication in the New England Journal of Medicine (NEJM). In a recent Letter to the Editor in the journal, our DC investigators report the case of a patient seen at the San Francisco General Hospital with symptoms of autoimmune polyglandular syndrome type 1 (APS1).

APS1 is a disease characterized by multiple autoimmune disorders, often including type 1 diabetes. As a leader in research on the AIRE gene and APS1, Mark Anderson, MD, PhD, the Robert B. Friend and Michelle M. Friend Endowed Chair in Diabetes Research, was consulted on this case. At first it appeared that this patient had no defect in the AIRE gene that is normally associated with APS1. However, the UCSF team discovered a thymic tumor that showed a defect in AIRE function. Because AIRE normally prevents autoimmune disease by promoting deletion of T cells that recognize self-tissues, the lack of AIRE function in the thymic tumor caused the APS1. Though the development of autoimmunity is complex, the discovery of a molecular link between two uncommon causes of autoimmunity--thymic tumors and APS1--has implications for treating all types of autoimmune disease. It also highlights that acquired problems with the AIRE gene can provoke autoimmunity later in life. An APS1 and Autoimmune Disease Clinical Trial has been launched to further explore these issues of autoimmunity. For more information, contact Mickie Cheng, MD, PhD who also serves as first author on the NEJM publication.

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