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The Genetic Basis of Human Sjögren's Syndrome

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Lay Abstract

Essentially all traits, including susceptibility to disease, are influenced by inherited genetic variation. In Sjogren's syndrome (SS), multiple genes are certain to be involved, however, less than 1% of the estimated 25,000 human genes have ever been tested for a role in SS. As a result, the fundamental cause of this complex disorder remains unknown. Powerful tools are now available that allow researchers to comprehensively screen all genes for a potential role in disease. Our goal is to test essentially every gene for association with SS. We will identify those that contribute to development of disease and characterize the biological pathways that are altered in SS patients. Results from these studies are likely to reveal novel insights into SS that will be informative for all aspects of SS research. Understanding the fundamental cause of human SS will provide new opportunities for developing alternative approaches to diagnosing and treating SS.

Scientific Abstract

Sjogren's syndrome (SS) is a complex disorder influenced by both genetic and environmental factors. However, the genetics of human SS are virtually unexplored. Recent large-scale genetic studies in lupus, rheumatoid arthritis (RA), and other related diseases have pinpointed hundreds of important disease genes and revealed completely novel disease pathways. We propose to use powerful genome wide association approaches to screen >1.1 million genetic variants for association with SS. Significant associations will be confirmed through replication studies in independent subjects. This project is a large, multidisciplinary, international effort to discover the fundamental cause of human SS. Every gene and pathway established as causal in SS has significant potential for providing rapid advances in our understanding of disease mechanisms. These results will lay important groundwork for developing accurate diagnostic tools and identifying therapeutic targets tailored to disease relevant pathways in SS.