



INVITED LECTURE PRESENTATION

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GWAS in IMIDs

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Dysregulation of the immune system is a fundamental disease mechanism, yet is incompletely understood. Human genetics offers one approach to understand dysregulation of the immune system in human diseases. Until recently, a major bottleneck has been the identification of DNA variants (alleles) that contribute to risk of disease. Through international collaborations, many groups have employed genome-wide mapping approach on thousands of samples using common single nucleotide polymorphisms (SNPs). These studies, termed genome-wide association studies (GWAS), have discovered >150 loci that confer risk of common autoimmune diseases such rheumatoid arthritis (RA), type 1 diabetes (T1D), systemic lupus erythematosus (SLE), celiac disease, and inflammatory bowel disease (IBD). For most of these autoimmune diseases, these risk alleles explain approximately 25% of disease burden, whereas >50% of autoimmune disease risk is thought to be genetic, indicating that additional risk alleles remain to be discovered. In addition to finding new risk alleles, a next critical step is to understand how these alleles disrupt normal immune function. In this way, the field of human genetics hopes to gain insight into fundamental mechanisms that lead to autoimmunity, which in turn could help guide the development of novel therapies.

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