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Genome editing to define the function of risk loci and variants in rheumatic disease

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Abstract

Discoveries in human genetic studies have revolutionized our understanding of complex rheumatic and autoimmune diseases, including the identification of hundreds of genetic loci and single nucleotide polymorphisms that potentially predispose individuals to disease. However, in most cases, the exact disease-causing variants and their mechanisms of action remain unresolved. Functional follow-up of these findings is most challenging for genomic variants that are in non-coding genomic regions, where the large majority of common disease-associated variants are located, and/or that probably affect disease progression via cell type-specific gene regulation. To deliver on the therapeutic promise of human genetic studies, defining the mechanisms of action of these alleles is essential. Genome editing technology, such as CRISPR–Cas, has created a vast toolbox for targeted genetic and epigenetic modifications that presents unprecedented opportunities to decipher disease-causing loci, genes and variants in autoimmunity. In this Review, we discuss the past 5–10 years of progress in resolving the mechanisms underlying rheumatic disease-associated alleles, with an emphasis on how genomic editing techniques can enable targeted dissection and mechanistic studies of causal autoimmune risk variants.

Biograph :

Soumya Raychaudhuri is a Professor of Medicine and Biomedical Informatics at Harvard Medical School, and an Institute Member at Broad Institute. He is the JS Coblyn and MB Brenner Distinguished Chair in Rheumatology/Immunology and a practicing rheumatologist at Brigham and Women's Hospital. He is the Director for the Center for Data Sciences at Brigham and Harvard. In addition, he serves as a Visiting Professor in Genetics at the University of Manchester. His research focuses on human genetics and computational genomics to understand immune-mediated diseases.