Recent advances in the genetics of rheumatoid arthritis

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ABSTRACT

Family studies have long demonstrated that rheumatoid arthritis susceptibility is linked to an underlying genetic component, both inside and outside of the major histocompatibility complex (MHC). The last decade has seen incredible advances in the discovery of risk loci for rheumatoid arthritis; now there are about 100 disease loci that have been identified. However, understanding the link between genetic loci and disease mechanism, is contingent on investigators identifying causal alleles and elucidating how they function to modify disease susceptibility. We are now just beginning to make this key step. Here we present recent work on (1) efforts to identify rheumatoid arthritis loci outside of the MHC region, (2) efforts within our own group to localize MHC effects to functional amino acid sites within HLA genes, separately for seronegative and seropositive rheumatoid arthritis, and (3) methodological advance to connect non-MHC loci to functional alleles that influence gene regulation in specific immune cell-types.
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RESEARCH TOPICS
The focus of my research is in computational biology, human genetics, and rheumatology. I have developed analytical methods to interpret large genomic data sets, including gene expression data, biological text, and data generated by human genetic studies. I have contributed to advancing human genetics by discovering key pathogenic loci in rheumatoid arthritis, cancer, age-related macular degeneration, and multiple sclerosis.

EDUCATION
1997 B.S., B.A. (summa cum laude), Biophysics, Mathematics, State University of New York at Buffalo, Buffalo, NY
2002 Ph.D., Biomedical Informatics, Stanford University, Palo Alto, CA
2004 M.D., Medicine, Stanford University

POSITIONS
2011.4– Assistant Professor, Medicine, Harvard Medical School
2012.8– Professor in Genetics (part-time), Institute of Inflammation and Repair, Faculty of Medical and Human Sciences, University of Manchester, Manchester, UK

AWARDS (SELECTED)
2012 William F. Milton Fund Award, Harvard University Cambridge, MA
2012 Philip S. Magaram, Esq. Research Award, Arthritis Foundation
2013 Doris Duke Foundation Clinical Scientist Development Award