Rheumatologists advance genetic research related to disabling form of arthritis

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John D. Reveille, M.D., is the principal investigator of a new study on a disabling form of arthritis. Credit: The University of Texas Health Science Center at Houston

Work done in part by researchers at The University of Texas Health Science Center at Houston has led to the discovery of two new genes that are implicated in ankylosing spondylitis (AS), an inflammatory and potentially disabling disease. In addition, the international research team pinpointed two areas along stretches of DNA that play an important role in regulating gene activity associated with the arthritic condition.

The findings, a critical milestone in the understanding of AS, are published in the January issue of Nature Genetics, a journal that emphasizes research on the genetic basis for common and complex

diseases. "This helps us better understand what is driving this disease and gives us direction for new treatments and diagnostic tests," said John D. Reveille, M.D., the study's principal investigator and professor and director of the Division of Rheumatology and Clinical Immunogenetics at The University of Texas Medical School at Houston.

Reveille, the university's Linda and Ronny Finger Foundation Distinguished Chair in Neuroimmunologic Disorders, and Matthew A. Brown, M.D., professor of immunogenetics at Australia's University of Queensland, led the research by the Triple "A" Spondylitis Consortium Genetic Study (i.e. the TASC or Australo-Anglo-American Spondylitis Consortium). Based on work from a genome-wide association scan, the team identified genes ANTXR2 and IL1R2 as well as two gene deserts, segments of DNA between genes on chromosomes 2 and 21 that are associated with ankylosing spondylitis. Importantly, the study also confirmed the Triple "A" Australo-Anglo-American Spondylitis Consortium's previously reported associations of genes IL23R and ERAP1, formerly known as ARTS1.

Reveille, chief of rheumatology at Memorial Hermann-Texas Medical Center, said the genetic discoveries bring the scientific community closer to fully understanding AS, a chronic form of arthritis that attacks the spine and also can target other joints and organs in the body. The Centers for Disease Control and Prevention for the National Arthritis Data Workgroup estimates that AS and its related diseases affect as many as 2.4 million people in the United States. It generally strikes patients in their teens, 20s or 30s and can cause a complete fusion of the spine, leaving patients unable to straighten and bend.

Steve Haskew, who has lived with AS for more than three decades, said these genetic discoveries offer hope to patients, especially those newly diagnosed.

"When I first started experiencing lower back pain and the aching joints, no one could tell me what was wrong," said Haskew, co-leader of an AS support group. "It's fascinating to see how far we've come and how much has been learned about the disease."

Laurie Savage, co-principal investigator and executive director of the Spondylitis Association of America (SAA) said, "These new breakthroughs are, indeed, good news for those whom we serve. It is very encouraging to know that the health impact and economic consequences of spondyloarthritis in the world eventually will be contained as a direct consequence of the dedication of Drs. Reveille, Brown and colleagues, and that of the many individuals affected by spondyloarthritis who have participated in these studies."

More information: "Genomewide association study of ankylosing spondylitis identifies multiple non-MHC susceptibility loci," *Nature Genetics,* January 10.

Provided by University of Texas Health Science Center at Houston