

For the Alliance for Lupus Research
Contact: Sam Rogers, 646.884.6091

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Steps toward Stopping Autoimmune Disease: International consortium identifies genes linked to lupus

NEW YORK, January 20, 2008—A landmark genetic study has identified multiple genes linked to systemic lupus erythematosus (SLE), or lupus, a debilitating autoimmune disease that affects an estimated 1.4 million Americans.

Lupus can affect the joints, kidneys, heart, lungs, brain and blood and occurs in about 31 out of every 100,000 people. Women are nine times more likely than men to develop the condition, which is often difficult to diagnose.

In 2005 the Alliance for Lupus Research (ALR) formed and supported the International Systemic Lupus Erythematosus Genetics (SLEGEN) Consortium, charging scientists with searching for genetic variants that might predispose an individual to developing lupus.

Published in the January 20, 2008, issue of *Nature Genetics*, initial study results uncovered several genes linked to lupus and underscore the importance of genetic variants in diseases that affect immune function. The findings will ultimately lead to new therapies and earlier diagnosis.

“The SLEGEN study is a model for collaborative genetic research,” said Mary Kuntz Crow, M.D., an immunity and inflammation specialist at the Hospital for Special Surgery in New York and 2008 chair of the ALR Scientific Advisory Board. “The ALR approach of supporting investigations targeted toward developing new therapies for people with lupus is unique and meaningful.”

Project co-Director Carl Langefeld, Ph.D., added, “These results suggest biologic pathways that help us understand the condition better and suggest additional genetic and non-genetic triggers. In addition, they will help delineate the genetic distinctions between rheumatoid arthritis, lupus and other autoimmune diseases, which could lead to earlier, more accurate diagnoses.”

Langefeld, who is director of the Center for Public Health Genomics at Wake Forest University in Winston-Salem, N.C., also noted how satisfying the study results were. “This is one of those things that, at the end of your career, you can look back on and smile because you believe it will make a real difference.”

SLEGEN Director John B. Harley, M.D., Ph.D., explained that the study found strong evidence of association with multiple single nucleotide polymorphisms (SNPs) in three genes: ITGAM; KIAA1542; PDK; and at SNP rs10798269, a DNA unit not found within any known gene. SNPs are chromosome locations where a single unit of DNA may vary from one person to another.

The results also showed evidence linking lupus to nine other genes. "I would have been satisfied with finding one gene. The fact that we present 13 strong candidates, supported by data that are 99% accurate is tremendous," added Harley, who heads the Arthritis & Immunology Research Program at the Oklahoma Medical Research Foundation in Oklahoma City.

Researchers studied the DNA of more than 6,700 women, including individuals with lupus, their family members and control subjects. Harley said they scanned the entire genome for more than 317,000 SNPs with the goal of identifying SNPs linked to lupus.

"SLEGEN's purpose is to do the genomics," Harley said. "The mechanism of the disease by which these genes cause lupus will be seized upon by scientists who are expert in those pathways to develop new strategies for prevention and therapy and reduce the burden of suffering this disease causes."

Langefeld said the accomplishments made in the SLEGEN research wouldn't have been possible without the support of ALR. "We simply would not have been able to do it without them."

"We are very pleased to see the findings in print and are grateful that the ALR support for SLEGEN has had such a fruitful outcome," said Joseph E. Craft, M.D., chief of rheumatology at Yale University School of Medicine and former chair of the ALR Scientific Advisory Board.

The Alliance for Lupus Research (ALR) is a national voluntary health organization based in New York City and founded in 1999. Chaired by Robert Wood Johnson IV, it has become the world's largest private source of lupus research funds and has committed \$50 million to lupus-specific research projects since its inception. The organization's mission is to find better treatments and ultimately prevent and cure lupus by supporting medical research. For more information, visit www.lupusresearch.org.

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