

# **Making precision medicine a reality: Genomics researchers unveil road map to disease origin**

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Understanding the genetic and biologic basis of diseases like rheumatoid arthritis, cancer, diabetes and Alzheimer's will lead to new therapies. Credit: University of Arizona Health Sciences

Researchers are one step closer to understanding the genetic and biological basis of diseases like cancer, diabetes, Alzheimer's and rheumatoid arthritis - and identifying new drug targets and therapies - thanks to work by three computational biology research teams from the University of Arizona Health Sciences, University of Pennsylvania and Vanderbilt University.

The researchers' findings - a method demonstrating that independent DNA variants linked to a disease share similar biological properties - were published online in the April 27 edition of npj Genomic Medicine.

"The discovery of these shared properties offer the opportunity to broaden our understanding of the [biological basis](#) of disease and identify new therapeutic targets," said Yves A. Lussier, MD, FACMI, lead and senior corresponding author of the study and UAHS associate vice president for [health sciences](#) and director of the UAHS Center for Biomedical Informatics and Biostatistics (CB2).

The researchers are striving to better understand the common genetic and biological backgrounds that make certain people susceptible to the same disease. They have developed a method to demonstrate how individual, disease-associated DNA variants share similar biological properties that provide a road map for disease origin.

Over the last ten years, genetics researchers have conducted large studies, called Genome Wide Association Studies (GWAS), which analyze DNA variants across thousands of human genomes to identify those that are more frequent in people with a disease. However, the impact of many of these disease-associated variants on the function and regulation of genes remains elusive, making clinical interpretation difficult.

A method to explore the biological impact of these variants and how

they are linked to disease was developed through the collaboration of bioinformatics and systems biology researchers Dr. Lussier; Haiquan Li, PhD, research associate professor and director for translational bioinformatics, Department of Medicine, UA College of Medicine - Tucson; Ikbel Achour, PhD, director for precision health, CB2; Jason H. Moore, PhD, director, Institute for Biomedical Informatics, Perelman School of Medicine, University of Pennsylvania; and Joshua C. Denny, MD, MS, FACMI, associate professor of [biomedical informatics](#) and medicine, Vanderbilt University, along with their teams.

In their new paper, the researchers demonstrate that DNA risk variants can affect biological activities such as gene expression and cellular machinery, which together provide a more comprehensive picture of disease biology. When DNA risk variants for a given disease were analyzed in combination, similar biological activities were discovered, suggesting that distinct risk variants can affect the same or shared biological functions and thus cause the same disease. More detailed analyses of variants linked to bladder cancer, Alzheimer's disease and [rheumatoid arthritis](#) showed that two variants can contribute to disease independently, but also interact genetically. Therefore, the precise combination of DNA variants of a patient may work to increase or decrease the relative risk of disease.

The team of researchers also is pursuing the development of methods to unveil the biological incidence of "long-time overlooked" DNA variants with the aim to more precisely inform clinical decisions with treatments tailored to a patient's genetic and biological background. Since two of these research teams (Lussier's and Denny's) [recently committed](#) to the White House Precision Medicine Initiative (PMI), this innovative study demonstrates how strategic collaboration is key to making precision medicine a reality, noted Dr. Lussier.

The paper, "Integrative genomics analyses unveil downstream biological

effectors of disease-specific polymorphisms buried in intergenic regions," has been identified as one of the best 30 of the year in computational biology and bioinformatics, and will be presented as a "highlight of the year" at the 2016 Intelligent Systems for Molecular Biology (ISMB) conference, the largest international conference of computational biology/bioinformatics, in July in Orlando, Fla.

**More information:** Haiquan Li et al, Integrative genomics analyses unveil downstream biological effectors of disease-specific polymorphisms buried in intergenic regions, *npj Genomic Medicine* (2016). [DOI: 10.1038/npjgenmed.2016.6](https://doi.org/10.1038/npjgenmed.2016.6)

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