Understanding and Improving Variant Interpretation in Minoritized and Underserved Populations

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Adam Gordon, PhD, is a member of the Center for Genetic Medicine. His laboratory investigates the implementation of genomic testing in medical and consumer contexts, and the process of variant interpretation that seeks to translate this testing into actionable clinical findings.

What are your research interests?

Our genomes are more accessible than ever before through commercial and clinical testing. However, the accumulation of this personal genomic data has significantly outpaced our ability to interpret it. My research focuses on how we decide what genetic variants mean, how we identify genetic variation that is clinically actionable and how to implement genomic medicine broadly and equitably without exacerbating the disparity, bias and scientific racism that's pervasive in medicine. As these questions are not unique to a specific clinical domain, I have active projects in oncology, cardiology, rheumatology, ophthalmology and other fields.

What is the ultimate goal of your research?

My research has three ultimate goals: 1) To refine our understanding of clinical actionability and penetrance in genomic medicine through data-driven approaches that integrate population and family-based cohorts of genomic and EHR data; 2) To broaden access and utility of genetic testing, especially in communities where such testing is beneficial but underutilized; and 3) To disrupt harmful notions about genetic determinism and scientific racism in medicine and the public at large through intersectional genomics education.

How did you become interested in this area of research?

In 2001, my biology teacher put up a poster of the map of the brand-new draft human genome. I was utterly fascinated at the idea that our genomes are what make us all fundamentally the same, but also fundamentally unique. That's when I knew I wanted to be a human geneticist.

My early work and interests were focused on population genetics and human evolution, thinking about the relationships between common and rare genetic variation, how these variants assort into different population groups, and how we decide which variants "do something." However, as I saw genomics expanding rapidly into medicine, I realized the extent to which concepts from human genetics have been misunderstood and misused in ways I believe are directly harmful to the public, and especially to minoritized communities. I believe those of us in medical research have a duty to acknowledge and repair these injustices, and I try to align my research with this intention.

What types of collaborations are you engaged in across campus (and beyond)?

As a team scientist, my goal is to partner with groups with deep expertise in specific clinical domains. Combining this expertise with emerging genomic technologies and data from large-scale population cohorts can lead to new insights into risk management, treatment and the biology of disease. Some current examples include studying the genetic architecture of sudden cardiac death with Greg Webster, MD, developing polygenic predictors of lupus risk with Theresa Walunas, PhD, and building a registry of individuals undergoing genetic testing for hereditary cancer risk with Mohammad Abbass, MD, PhD.

I have also for many years been a part of the national electronic Medical Records and Genomics (eMERGE) Network, which in its current iteration is studying the utility of polygenic risk testing in primary care: I am leading the prostate cancer arm of this study. Additionally, I serve on the American College of Medical Genetics secondary findings workgroup, using data to create policy about what genetic variation is actionable, and what sorts of incidental, actionable variation labs and providers have an ethical duty to report.

Finally, I'm very passionate about genomics education; I've worked with Nia Heard-Garris, MD, Susanna McColley, MD, and others to develop a course on anti-racist research strategies for current physicians, and recently completed a major revamp of the medical school genetics curriculum as a co-leader of the Foundations 1 module.

How is your research funded?

My research is primarily funded through the National Institutes of Health, especially the National Human Genome Research Institute, as well as the National Cancer Institute through Northwestern's Prostate Cancer SPORE. I also have some funding through foundations, particularly the American Heart Association.

Who are your mentors?

I am eternally grateful to those who have mentored me, and I carry their lessons with me every day. Specifically, my PhD advisor Debbie Nickerson and postdoc advisor Gail Jarvik; they taught me that the key to good science is teamwork, and that our responsibility as scientists is to be aspirational ...

Read more on page 8.