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Reducing ascertainment bias in Pharmacogenomic research

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Statement of the Problem: There is a well-established contribution of genetic variation to drug response that has resulted in the expectation of personalized optimization of drug efficacy and the minimization of drug toxicity. While the majority of drugs currently used in clinical practice lack companion genetic tests for therapeutic effects and/or adverse drug response avoidance, a variety of known genetic determinants of drug response (pharmacogenetics) have been documented and clinically validated. One of the challenges facing the comprehensive identification of pharmacogenetic (PGx) variants is the documented ascertainment bias in genomic research participation. Given that the uptake of PGx data in clinical care has been relatively slow, there is an opportunity to reduce this bias and increase the generalizability of results to clinical communities across the United States. The purpose of this study is to explore the impact of ascertainment bias on the translation of PGx research into clinical care. Findings: PGx panels such as the Affymetrix DMET array are missing important PGx variants that are rare in populations of European descent. Conclusion & Significance: There is an opportunity to expand the communities that participate in genomic and pharmacogenomic research, reduce sampling ascertainment bias, and increase the generalizability of genomic research findings to peoples living in the United States.

Biography

Laura Scheinfeldt is program manager and principal investigator of the National Institute of Neurological Disorders and Stroke (NINDS) Human Genetics Resource Center at the Coriell Institute for Medical Research. Her research has focused on the distribution of human genetic variation within and among populations living across the world and the relationship between genetic and phenotypic variation. Using an interdisciplinary approach, she has studied how genetic variation contributes to disease and drug response and has applied this research to precision medicine. For the past five years, she has worked with the Coriell Personalized Medicine Collaborative (CPMC) to understand how genetic and non-genetic factors impact health-related traits; she has worked on understanding how ascertainment biases in the current research literature impact marginalized populations and contribute to health disparities; and she has worked on identifying the subset of common genetic risk factors for complex disease that may motivate preventive health behaviors.

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