



JOSHUA C. DENNY, MD, MS

A JOURNEY TO PRECISION MEDICINE

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4:00 P.M.

208 LIGHT HALL



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Upcoming Discovery Lecture:

EMERY BROWN, M.D., PH.D.
Department of Brain and Cognitive Sciences

February 11, 2016
208 Light Hall / 4:00 P.M.

VANDERBILT  UNIVERSITY
MEDICAL CENTER

A JOURNEY TO PRECISION MEDICINE

Precision medicine offers the promise of improved diagnosis and for more effective, patient-specific therapies. The pursuit of precision (or personalized) medicine is as old as medicine itself but has dramatically accelerated in recent years. Typically, such studies have been pursued using research cohorts. At Vanderbilt, we have linked de-identified electronic health records (EHRs), to a DNA repository, called BioVU, with nearly 215,000 samples. Finding phenotypes in the EHR can be challenging, but the combination of billing data, laboratory data, medication exposures, and natural language processing has enabled efficient study of genomic and pharmacogenomic phenotypes using real-world data. The deep phenotype information in the EHR also enables the inverse experiment – starting with a genotype and systematically surveying all the clinical phenotypes with which it is associated – the phenome-wide association study (PheWAS). PheWAS has highlighted the influence of genetics across many phenotypes, to give us a better understanding of the real-world implications of genomic variants. PheWAS has other exciting capabilities, unforeseen at the time of its development, including unforeseen capabilities such as discerning the contribution of Neanderthal interbreeding to human disease, enabling subtyping of disease, and repurposing medications. The paradigm of EHR-based clinical and genomic research form a key enabling platform for the new NIH Precision Medicine Initiative (PMI), which will seek to engage at least 1 million individuals across the US. The PMI will redefine our understanding of disease onset and progression, treatment response, and health outcomes through measurement of molecular, environmental, and behavioral factors that contribute to health and disease. The ultimate goals include more accurate diagnoses, more rational disease prevention strategies, better treatment selection, and the development of novel therapies.



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Dr. Joshua Denny is an Associate Professor in the Departments of Biomedical Informatics and Medicine at Vanderbilt University Medical Center. He completed an internal medicine residency as a Tinsley Harrison Scholar at Vanderbilt. His interest in medical informatics began with development of natural language processing systems applied to clinical text. He currently focuses on applications of algorithms to accurately identify phenotypes from electronic medical record data, and using the electronic medical record to discover genome-phenome associations to better understand disease and drug response. Through this, he first described the phenome-wide association study (PheWAS) method applied to electronic medical record data. He also helps lead efforts for local and network pharmacogenetics implementation activities. He is part of the Electronic Medical Records and Genomics (eMERGE), Pharmacogenomics Research Network (PGRN), and the Implementing Genomics in Practice (IGNITE) networks. He is past recipient of the American Medical Informatics Association New Investigator and Homer Warner awards. Dr. Denny also remains active in clinical care and in teaching medical students.
