JOSHUA C. DENNY, MD, MS
A JOURNEY TO PRECISION MEDICINE

FEBRUARY 4, 2016
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.
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.