

The eMERGE Network: A national consortium of electronic health record-linked biobanks furthering large-scale genetic research *W.A. Wolf¹, R.L. Chisholm¹, C.G. Chute², G. Jarvik³, E. Larson³, D.R. Masys⁴, C.A. McCarty⁵, D.M. Roden⁴, J.P. Struewing⁶*. 1) Northwestern University, Chicago, IL; 2) Mayo Clinic, Rochester, MN; 3) Group Health Cooperative/University of Washington, Seattle, WA; 4) Vanderbilt University, Nashville, TN; 5) Marshfield Clinic Research Foundation, Marshfield, WI; 6) National Human Genome Research Institute, Bethesda, MD.

Medical research institutions are developing biorepositories of genomic DNA coupled to electronic health record (EHR) information generated by routine clinical care. The goal of the Electronic Medical Records and Genomics (eMERGE) Network (www.gwas.net), organized by NHGRI in late 2007, is to investigate how such resources can be leveraged for genome and informatics science, with extensive ELSI input. Each of the five participating sites will use natural language processing or other tools to identify cases with defined phenotypes and controls for genome wide analyses in ~15,000 subjects, and will share data with the scientific community through NIH's dbGaP. The target phenotypes include asthma, type II diabetes, low HDL, cataracts, myocardial infarction, peripheral arterial and carotid disease, normal QRS duration, late life dementias (Alzheimer's disease), and statin adverse events. Initial studies underway include testing computational algorithms to identify subjects meeting phenotype criteria, comparing these tools across diverse EHR systems, assessing the potential for combining cases or controls from different sites, and initiation of mechanisms to optimize community consultation. The goal of these studies is to contribute to our understanding of disease and develop recommendations to improve the utility of EHRs for research, setting the stage to integrate genomic and EHR data to achieve the vision and inform best practices for personalized medicine.