

Table S1. Summary of eMERGE-II Activities at Each Site

Institution	Aim 1	Aim 2	Aim 3	Aim 4	Aim 5
Cincinnati Children's Hospital and Boston Children's Hospital	Demonstrate real-time execution of phenotypic selection across two distinct pediatric institutions with disparate EMR systems as a model for ensuring phenotypic standardization and for national scalability	Contribute to phenotype selection and perform GWAS and PheWAS	Use return of <i>COMT</i> and <i>CYP2D6</i> research results to explore parents' responses to and use of their children's results and understand factors that influence their decisions about learning incidental findings	Explore clinicians' perceptions of pharmacogenetic research results after EMR integration	
Children's Hospital of Philadelphia	Adapt eMERGE phenotype algorithms relevant to pediatric cohorts with circulating lipid levels as the primary phenotype	Define novel phenotypes with relevance to adult and pediatric cohorts (asthma, ADHD, atopic dermatitis, and GERD)	Conduct GWAS with minimal risks to patient privacy from sharing of EMR data	Develop consent and community consultation procedures for conducting research	Begin incorporating genomic research results into clinical care
Geisinger Clinic	Identify genetic variants associated with AAA, extreme obesity, and related conditions	Incorporate genomic data into clinical care, using EMR-based CDS	Identify sociocultural concerns of patients in rural areas regarding return of genetic findings. Provide patient and physician education		
Group Health	Study infectious disease susceptibility (BuGWAS), specifically susceptibility to <i>Clostridium difficile</i> , reactivation of the varicella zoster virus, and fungal nail infection (onychomycosis)	Investigate the relationship of chromosomal abnormalities (ChroWAS) identified by GWAS with bone marrow disorders	Assess challenges of integrating genomic data into clinical care: interview medical system leaders, focus groups with physicians and patients, and develop and test prototype	Collaborate within and extend eMERGE through exchange of knowledge, technology, and best practices for generating EMR-based GWAS and integrating with clinical care	
Marshfield Clinic Research Foundation	Develop and validate electronic algorithms and efficacy of medical therapy for ophthalmic conditions	Undertake genetic discoveries for ophthalmic conditions and pharmacogenetics	Consultat with the general community and clinicians related to the incorporation of GWAS results into EMR		
Mayo Clinic	Identify genetic variants for inter-individual variation in cardio-respiratory fitness, and susceptibility to VTE and CHF using validated and transportable	Quantify genetic risk of CHD and statin myopathy. Develop risk communication tools with clinical and genetic components to both patients and care	Develop informatics approaches to incorporate genomic information and relevant clinical decision support tools into the EMR	Conduct a randomized-clinical trial to investigate how patients respond to genotype-informed CHD risk	

Mount Sinai School of Medicine	phenotyping algorithms Develop and implement secure prototype Biobank-EMR genomic data interface and EMR-enabled genomic CDS in clinical care	providers Develop phenotyping algorithms for chronic kidney disease and its progression	Expand GWAS for cardiovascular and renal phenotypes across minority populations	Implement novel solutions incorporating genomic results with EMR	Explore innovative approaches for community-participatory education and research in genomic medicine
Northwestern University	Expand development of phenotype algorithms and detection of new genomic associations	Consult with physicians and patients regarding the utility of real world examples of clinically relevant genotypes and to inform clinical activities	Develop technical approaches for integrating genetic variant data into the EMR	Evaluate impact of key translational elements (e.g. regulatory barriers, EMR CDS tools, and provider and patient education) and disseminate lessons learned and best practice recommendations	
Vanderbilt University	Accelerate development of phenotype algorithms	Identify combinations of genotypes highly predictive of disease or drug response outcomes	Study patient perspectives of genetic testing to guide drug prescribing	Develop tools to maximize sharing of genomic information while preserving privacy	
Coordinating Center Vanderbilt University with subcontract to the Pennsylvania State University	Facilitate communication between eMERGE sites and coordinate in-person Steering Committee meetings	Provide project management and administrative support for all workgroups and ESP	Provide leadership in advancement of electronic phenotyping, genomic data management, and data privacy	Develop and maintain web portal (www.gwas.org)	Provide organizational dashboards with site-specific and network progress towards goals

AAA, abdominal aortic aneurysm; ADHD, attention deficit hyperactivity disorder; CDS, Clinical Decision Support; CHD, coronary heart disease; CHF, chronic heart failure; EMR, electronic medical records; ESP, external scientific panel; GERD, gastroesophageal reflux disease; GWAS, genome-wide association study; PheWAS, phenome-wide association study; VTE, venous thromboembolism.
For details on biorepositories and EMR, see Table 1 in the main section.