Electronic medical record

The Electronic medical record and genomics (eMERGE) network review1

**NHGRI, March 2007, Phase I [eMERGE-I]**

Five sites:

“In September 2007, grants were awarded to five sites (hereaf- ter referred to as eMERGE-I)—Group Health Cooperative/University of Washington, Marshfield Clinic, Mayo Clinic, Northwestern University, and **Vanderbilt University**, which also served as the network’s **coordinating center**.”  
eMERGE-II (9 sites):

CHOP [Children’s hospital of Philadelphia], GHC & U Wash, Mount Sinai, CCHMC & BCH [Cincinnati Children’s Hospital Medical Center with Boston Children’s Hospital], Marshfield & Essentia, Northwestern, Geiginger, Mayo, Vanderbilt

Three major aims:

1 EMR for robust electronic phenotyping;

2 conduct GWAS using the phenotypes derived in the above-mentioned first aim; PheGWAS2

3 explore the ethical, legal, and social implications associated with EMR-based GWAS and wide-scale data sharing.

The workgroups included:  
an informatics group [published a phenotyping algorithm, PheKb: Phenotype KnowledgeBase, 20http://www.PheKb.org]. Developed by the first site and deployed to the second, and then across the network.   
a genomics group [imputation, strand issue, relatedness, ],   
and a consent and community consultation group [model language for EMR-linked ].

1. Gottesman, O. *et al.* The Electronic Medical Records and Genomics (eMERGE) Network: past, present, and future. *Genet. Med.* **15**, 761–71 (2013).

2. Denny, J. C. *et al.* PheWAS: Demonstrating the feasibility of a phenome-wide scan to discover gene-disease associations. *Bioinformatics* **26**, 1205–1210 (2010).

3. Fullerton, S. M. *et al.* Return of individual research results from genome-wide association studies: Experience of the Electronic Medical Records and Genomics (eMERGE) Network. *Genet. Med.* **14**, 424–431 (2012).

**Lesson learned from Phase I**

1 Phenotyping across the network rather than local sites.

2 Most eMERGE Participants have consented to contributing their data to health research of any kind. Through network-wide projects, eMERGE-I was compelled to develop best practices for sharing genomic data and EMR-derived phenotypes while protecting the privacy of participants.

3 Returing of results (RoR) 3

**Phase II [eMERGE-II]**

Phenotyping workgroup

Genomics workgroup

RoR workgroup

Consent, education, regulation, and consultation workgroup: evaluating the impact of returning hemochromatosis results

EMR integration workgroup: PGx [pharmcogenomic] pilot project

Collaboration with external groups

Lessons

**1 Portability of electronic phenotypes within and outside eMERGE**

“There is currently no formal phenotyping language for the purpose of building EMR phenotyping algorithms nor is their a common approach to their implementation.

**2 Approaches to EMR integration of genomic information**

**3 Integration of pediatric sites**

**4 Longitudinal cost-effective genomic medicine discovery and implementation**

**5 Generalized framework for the return of genomic results**

**6 The eMERGE network in the context of a translational framework.   
T0: early phases focusing of biologic discoveries (eMERGE-I focused large on T0)  
T1: development of candidate health applications (eMERGE-II)**

**T2: assessing outcomes of intervertions**

**T3: move genomic findings into health practice.**

**T4: public health surveilance**