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 ].

**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. Turner syndrome, Klinefelter syndrome, FVL, hereditary hemochromatosis. There is no clear agreement how the finding should be returned. In some case, the clinically actionable results identified in EMR was already known. “In some case, health records shed light on ambiguous findings, increasing the likelihood that some findings represented acquired rather than congenital genetic changes.”

**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 surveillance**

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).