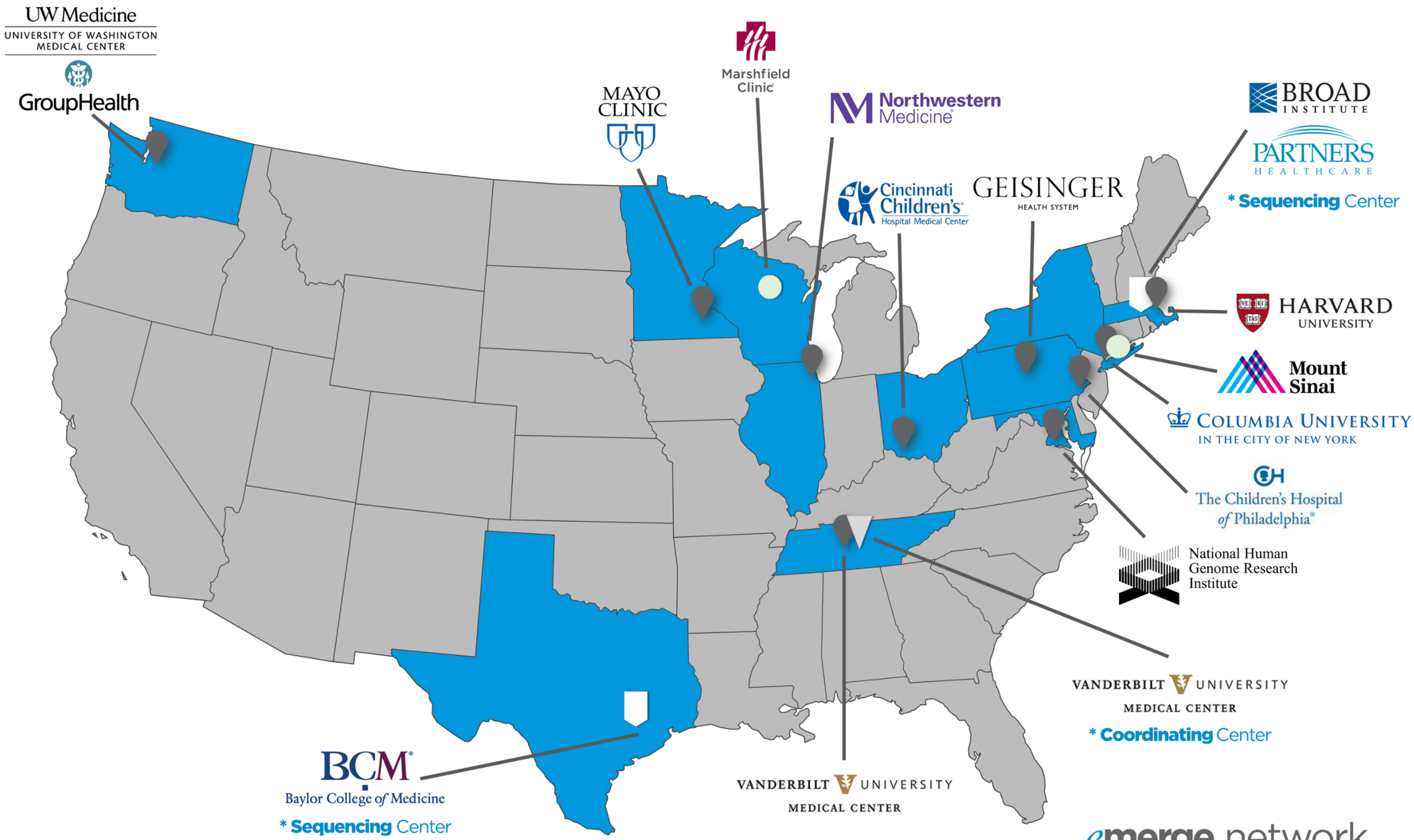
TOP 5 Consortium-wide Achievements

February 2, 2017



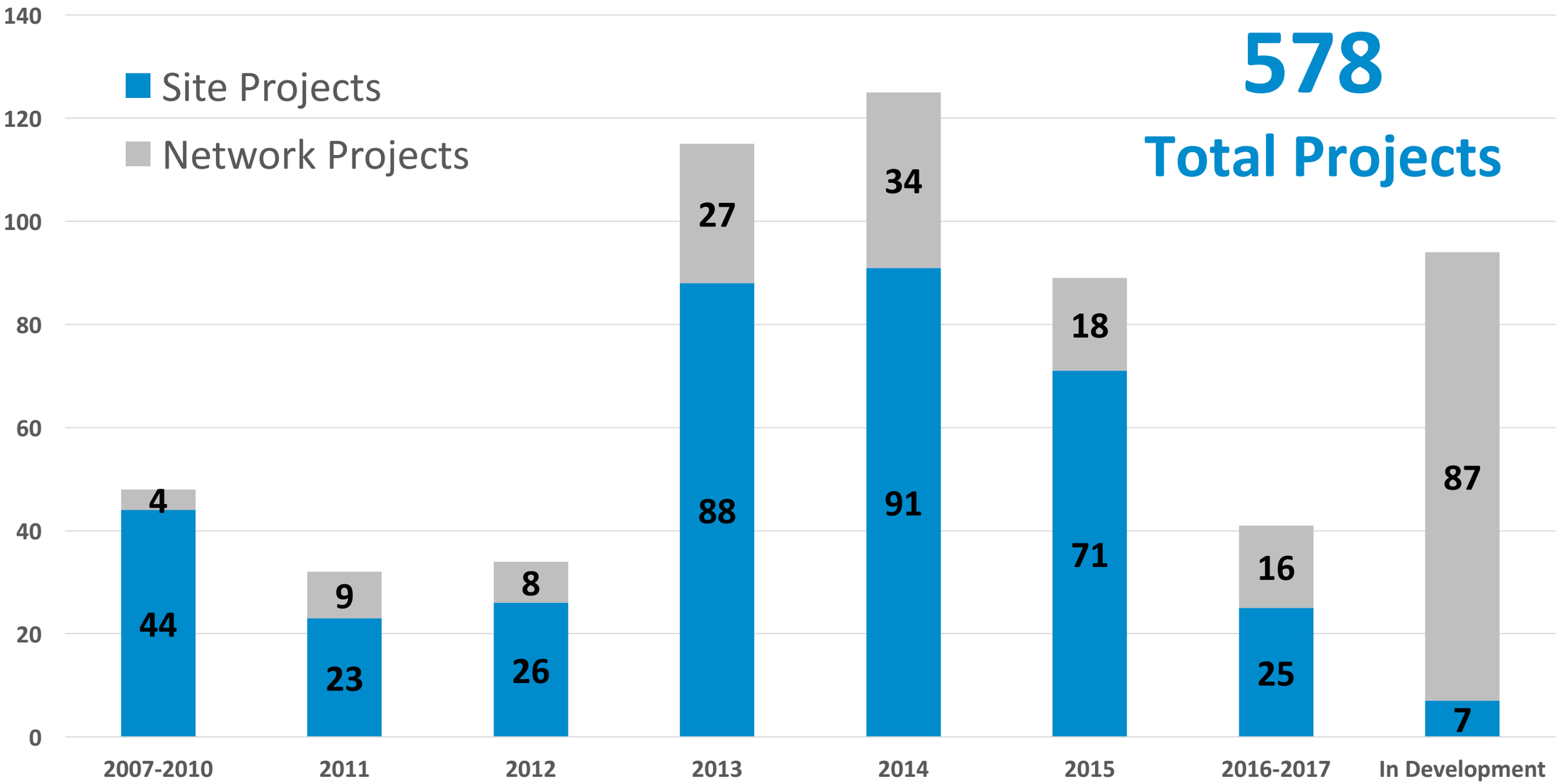
*e*MERGE III: What Do We Do?

SPECIFIC AIMS *of the* eMERGE Network

1. Sequence and assess clinically relevant genes presumed to affect gene function in about 25,000 individuals
2. Assess the phenotypic implications of these variants
3. Integrate genetic variants into EMRs for clinical care
4. Create community resources

*e*MERGE Publications *2007-2017*

Cumulative Citation Count: 15,985



High Throughput Phenotyping & PheKB

- Demonstrated that electronic health records can be used to define phenotypes that can be used in both discovery and implementation for Genomic Medicine

- PheKB (**Phe**notype **K**nowledge**B**ase)
  - Collaborative environment to building and validating electronic algorithms
  - Computational algorithm library
    - 37 finalized, public phenotypes
  - https://phekb.org/

- Tools and process allowed for computational and algorithm development cross collaboration around the world

PheKB a knowledgebase for discovering phenotypes from electronic medical records

Kirby JC, Speltz P, Rasmussen LV, Basford M, Gottesman O, et al. *PheKB: a catalog and workflow for creating electronic phenotype algorithms for transportability.* J Am Med Inform Assoc. 2016 Nov;23(6):1046-1052. doi: 10.1093/jamia/ocv202. **PMID: 27026615 PMCID: PMC5070514.**

100k Participant Genomic Dataset

- Data on over 100,000 participants and informatics tools with which to harness the data
  - Array data; PGRNSeq; eMERGE-Seq panel (coming soon, will add 25k)
- eMERGE Record Counter
  - Drag and drop demographics, phenotypes, ICD codes to obtain preliminary cohort counts
  - https://biovu.vanderbilt.edu/EmergeRC/
- SPHINX (**S**equence and **PH**enotype **IN**tegration E**X**change)
  - Search catalog by genes, drugs, and pathways
  - For each gene view: SNVs, pathways, drug interactions
  - For each variant view: SNPid, category, frequencies
  - https://www.emergesphinx.org/

Rasmussen-Torvik LJ, Stallings SC, Gordon AS, Almoguera B, Basford MA, et al. *Design and Anticipated Outcomes of the eMERGE-PGx Project: A Multicenter Pilot for Preemptive Pharmacogenomics in Electronic Health Record Systems*. Clinical Pharmacology & Therapeutics. 2014 Oct; 96(4):482-9. **PMID: 24960519 PMCID: PMC4169732**

emerge recordcounter

SPHINX
A resource of the *e*MERGE Network

eMERGE Pharmacogenomics (PGx)

- Multi-site test of the concept that genetic sequence information can be coupled to electronic medical records (EMRs) for use in healthcare.
- Genetic sequencing on a 9000 participant data set
- 82 pharmacogenetic genes investigated
- Many more opportunities for research on these data
  - PGx SNVs on the eMERGE-Seq panel
- Sites continue to collect utilization and outcomes data

https://emerge.mc.vanderbilt.edu/projects/emerge-pgx/

eMERGE PGx
A multi-center pilot of pharmacogenetic sequencing in clinical practice

Bush WS, Crosslin DR, Owusu-Obeng A, Wallace J, Almoguera B. et al. *Genetic variation among 82 pharmacogenes: The PGRNseq data from the eMERGE network. Clin Pharmacol Ther. 2016 Aug;100(2):160-9. doi: 10.1002/cpt.350.* ***PMID: 26857349 PMCID: PMC5010878.***

eMERGE PheWAS

- Phenome-wide association studies (PheWAS) analyze many phenotypes compared to a single gene-disease association.
  - 3144 SNPS present in NHGRI catalog (2012) in 13,835 individuals across 5 sites. 1358 phenotypes analyzed for each SNP.
  - Neanderthal PheWAS catalogue
  - https://phewascatalog.org/
- Developed methods for large scale genotype/phenotype analyses and implemented them across an entire collaborative Network

PheWAS Resources

Denny JC, Bastarache L, Ritchie MD et al. *Systematic comparison of phenome-wide association study of electronic medical record data and genome-wide association study data.* Nat Biotechnol. 2013 Dec;31(12):1102-10. **PMID: 24270849 PMCID: PMC3969265**

Integration *of* Genomic Data *into* EHRs to inform clinical care

- Infrastructure and tools, in particular decision support tools, to enable genomic medicine
- InfoButton
  - Explored use of infobuttons as a decision support tool to provide context specific links within the electronic health record (EHR) to relevant genomic medicine content
  - Assessed the coverage of content topics among information resources developed

(Overby CL, Rasmussen LV, Hartzler A, Connolly JJ, Peterson JF, et al. *A Template for Authoring and Adapting Genomic Medicine Content in the eMERGE Infobutton Project*. AMIA Annu Symp Proc. 2014 Nov 14;2014:944-53. **PMID: 25954402 PMCID: PMC4419923.**)

- CDS_KB (**C**linical **D**ecision **S**upport **K**nowledge**B**ase)
  - Partnership with IGNITE network
  - Goal is to catalog and share CDS implementation artifacts and design considerations for genomic medicine programs from a broad community of institutions
  - https://cdskb.org/

CDS_KB
CLINICAL DECISION SUPPORT KNOWLEDGEBASE