EMR and Genomic Discovery -- Beyond GWAS, Pharmacogenomics

Discussion Points

Issues for Discussion

- Discovery research should remain a high priority for eMERGE-III
  - Prioritize traits/phenotypes (disease associations and drug responses) for discovery research
  - More comprehensive analyses with existing data *vs* denser data generation (NGS, exon arrays)

- Address the importance of rare (but collectively common) functional variants
  - Implement appropriate genotyping/sequencing platforms to capture them
  - Develop analytical tool to detect their effects

Issues for Discussion

- Consider alternative study designs for discovery
  - Extreme discordant phenotype coupled with WGS, focusing on the tails of phenotypic distributions

- Expand data collection processes
  - Additional sources of DNA/RNA for genomic analyses
  - Capture data on environmental factors, co-morbidities for GxE interactions