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| **eMERGE Network: Manuscript Concept Sheet** |
| **Reference Number** *(to be assigned by CC)* | NT341 |
| **Submission Date** | April 26, 2019 |
| **Project Title** | Genomic Information for Clinicians in the Electronic Health Record: Lessons Learned from ClinGen and eMERGE |
| **Tentative Lead Investigator** *(first author)* | Marc Williams |
| **Tentative Senior Author** *(last author)* | Casey Overby Taylor |
| **All Other Authors**  | Nephi Walton, Scott Goehringer, Bob Freimuth, Luke Rasmussen, Sandy Aronson, Guilherme Del Fiol |
| **Sites Participating** | Geisinger, Johns Hopkins, Mayo Clinic, Northwestern University, Partners HealthCare, University of Utah, other interested sites |
| **Background / Significance** | Genomic information is increasingly used in clinical care. Clinicians without genetic training consistently state they are unprepared to use genomic information to care for their patients. There is also concern about where to find reliable information to guide the use of genomic results. One of the goals of the NHGRI-funded Clinical Genome Resource (ClinGen) project is to create a genomic knowledge repository to support the use of genomic information in clinical care. In this paper we will review the approaches used to make ClinGen information more accessible to clinicians using the electronic health record (EHR) and the obstacles preventing full integration. This project is a collaboration between the ClinGen and eMERGE EHRI workgroups. |
| **Outline of Project** | * Review collective experiences from EHR integration work done at sites within eMERGE
* Evaluate ClinGen information resources
* Perform expert analysis and attain group consensus on opportunities to use the EHR to provide genomic information to clinicians
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| **Desired Data - Common Variables\*** *(Available from the CC)* | [ ] Demographics [ ] ICD9/10 codes[ ] CPT codes[ ] Phecodes[ ] BMI | [ ] Common Variable Labs[ ] Common Variable Meds[ ] Other: Case/Control status on Phase I and [ ] Phase II phenotypes**(N/A)** |
| **Other Desired Data *(Available from participating sites)*** | Data will consist of narrative and/or written descriptions of experiences from eMERGE sites participating in EHRI efforts and ClinGen EHR WG. |
| **Desired Genetic Data** | [ ] eMERGE I-III Merged set (HRC imputed, GWAS)[ ] eMERGE PGx/PGRNseq data set [ ] eMERGEseq data set (Phase III)[ ] eMERGE Whole Genome sequencing data set[ ] eMERGE Exome chip data set[ ] eMERGE Whole Exome sequencing data set[ ] Other (not listed above): |
| **Does project pertain to an existing eMERGE Phenotype?** | [ ] Yes, if so please list [x] No |
| **Planned Statistical Analyses** | Summary statistics, descriptive analysis |
| **Ethical Considerations** | The project will be framed in a way that does not inappropriately expose protected IP of any EHR vendor. |
| **Target Journal** | Frontiers in Genetics Special Issue |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | May 15, 2019 – Project beginsJune 3, 2019 – First draft sent to co-authorsJune 17, 2019 – Co-authors return feedbackJune 24, 2019 – Second draft sent to co-authorsJune 28, 2019 – Co-authors return final feedbackJuly 1, 2019 – Submission to journal |

**\*Common Variables available across all datasets:**

* Demographics: sex, year of birth, decade of birth, race, ethnicity
* Codes: (repeated values & age at event): ICD, CPT, Phecodes
* BMI: (repeated value & age at event) height, weight, BMI
* Labs: (lab name, repeated lab value & age at event) Serum total cholesterol, LDL, HDL, Triglycerides, Glucose fasting/non-fasting/unknown, & White Blood Cell count
* Medications: (medication name, repeated, & age at event) Cerivastatin sodium, Rosuvastatin, Simvastatin, Fluvastatin, Pravastatin, Lovastatin, Atorvastatin, & Pitavastatin
* Other: Case/Control status on Phase I and Phase II phenotype: only on GWAS dataset participants