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| **eMERGE Network: External Collaborator Manuscript Concept Sheet** |
| **Reference Number** *(to be assigned by CC)* | NT301 |
| **Submission Date** | 8/16/2018 |
| **Project Title** | IGNITE Clinical Informatics Working Group: Genetic Data Pipeline Project |
| **Tentative Lead Investigator** *(first author)* | Paul Dexter (Indiana University) |
| **Tentative Senior Author** *(last author)* | Josh Peterson (josh.peterson@vanderbilt.edu)  |
| **eMERGE Site Sponsor & Contact** | Coordinating Center ; Josh Peterson (josh.peterson@vanderbilt.edu)  |
| **All Other Authors**  | Henry H. Ong; also specifically interested in eMERGE EHRi working group leads or designees. Other authors from IGNITE, CPIC, and CSER will be included. |
| **Sites Participating** | Any interested site from eMERGE, IGNITE, CPIC, or CSER |
| **Background / Significance** | As there are no turnkey solutions to integrate clinical genetic results into electronic health records, individual health systems have constructed unique and tailored infrastructures to manage clinical genetic results involving electronic health records and personal health records. The CIWG group of the IGNITE network has designed a survey and data collection instrument to describe the workflow and technical features (at the current point in time) of implemented clinical pipelines to transmit genetic data from a laboratory instrument (whether internal to the institution or external) to the point of care. |
| **Outline of Project** | We will request responses to a REDCap data collection sheet from IGNITE, eMERGE, CSER funded health systems and to interested participants representing other institutions involved with CPIC. We will summarize all responses and highlight common practices and challenges. No participant level data will be required. We request one representative from each site sign up. |
| **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 |
| **Other Desired Data *(Available from participating sites)*** | *Please specifically list out any data elements that participating sites would collect or extract from clinical or other sources for this project (i.e. not common variables above)* Participating sites will fill out a REDCap survey. |
| **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** | Descriptive statistics on data collection sheet responses |
| **Ethical Considerations** | None |
| **Available Funding or Resources** | This research is funded through IGNITE |
| **Target Journal** | TBD |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | 1. Complete data collection by 12/31/18
2. Submit manuscript by 3/1/19
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**\*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