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| **eMERGE Network: Manuscript Concept Sheet** |
| **Reference Number** *(to be assigned by CC)* | NT324 |
| **Submission Date** | January 17, 2019 |
| **Project Title** | Challenges in Returning Results in the eMERGE consortium |
| **Tentative Lead Investigator** *(first author)* | Colin M. Halverson |
| **Tentative Senior Author** *(last author)* | Ellen Wright Clayton |
| **All Other Authors**  | Georgia Wiesner, all other people who submit data and comment substantively on drafts |
| **Sites Participating** | eMERGE sites that submit data about challenges that we have faced |
| **Background / Significance** | It is critically important to understand the challenges that returning research results to participants entails both, if possible, to develop strategies to address challenges, and to understand the costs in time and resources required, which are critical to planning |
| **Outline of Project** | What we would like, if possible, are details about many times you encountered problems and what was required in terms of time and resources to address problems, with:   Deceased patients  Familial implications and RoR  Patients don’t open their results so they don’t remember that they got them back  Patients change their minds midstream about wanting the results back   Patients change their minds midstream about wanting results in EMR  Call back patients for retesting (mosaicism, germline, transplant, etc.)  Other types of mismatch between patient and sample  Can’t find patients  Can’t find clinicians to send the results for disclosure, including which clinicians you tried to reach  PCP refuses to return results  Reclassification/dubious variant calls  “Straight errors”Other unexpected difficulties If we missed something, please tell us by January 24, 2019 at which time we will finalize the list By January 31, please send information at minimum about how many of people in your cohort have experienced these problems. If you want to provide more details, that would be awesome. If we have data by January 31, we will have a draft mss. By mid-March (probably sooner) for comments.  |
| **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)*  |
| **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 |
| **Ethical Considerations** | It is critical that we understand and address the real world challenges that arise in returning results. |
| **Target Journal** | GiM |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | 1-31-2019 Receive information from co-authors on how many people in cohort have experience problems returning results. 3-15-2019 Draft manuscript completed |

**\*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