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| **eMERGE Network: Manuscript Concept Sheet** | | |
| **Reference Number**  *(to be assigned by CC)* | NT389 | |
| **Submission Date** | 4/27/2020 | |
| **Project Title** | Survey responses of participants who received negative results | |
| **Tentative Lead Investigator** *(first author)* | Hila Milo Rasouly, Julia Wynn | |
| **Tentative Senior Author**  *(last author)* | Georgia Wiesner, Ellen Wright Clayton | |
| **All Other Authors** | Representatives of sites who returned negative results and surveyed the recipients, Ingrid Holm, Kathy Zhao | |
| **Sites Participating** | Columbia (2 projects), Northwestern, UW/Kaiser, Mayo (2 sites), VUMC, CCHMC | |
| **Background / Significance** | The impact of receiving negative genomic screening results is incompletely understood. While many find these results reassuring, it is unclear how well their implications are understood, with whom their share their results, or how the process affected their view of testing. The eMERGE III project is analyzing the impact of receiving positive results in NT 349, 363, and 373. The eMERGE III project had 9 sites that returned negative results, a process analyzed in NT332, which found that almost 10,000 participants received negative results. Building on the experience at VUMC, which revealed some variation, we propose here to analyze the survey responses of those participants, which we anticipate will be one of the largest such groups to date and which would be an opportunity lost. | |
| **Outline of Project** | We will analyze the survey responses of participants who received negative results, using the same analytic approaches used for those who received positive results as well as those undertaken at VUMC of negative results and discuss how they add to our understanding of the impact of negative results. | |
| **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)*  Results from first participant 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  No | |
| **Planned Statistical Analyses** | Logistic regression for sites and demographics | |
| **Ethical Considerations** | None | |
| **Target Journal** | Genetics | |
| **Milestones**  *(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | We anticipate that this analysis can be rapid since the proposed first authors have created data dictionary to permit comparison among the surveys used at the sites and Kathy Zhao and Digna Velez Edwards analyzed VUMC survey results. If so, a manuscript could be completed within two months after completion of the analysis. | |

**\*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