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| **eMERGE Network: Manuscript Concept Sheet** |
| **Reference Number** *(to be assigned by CC)* | NT373 |
| **Submission Date** | 01/20/2020 |
| **Project Title** | **Psychologic impact on participants of receiving positive genomic results in eMERGE 3** |
| **Tentative Lead Investigator** *(first author)* | Ingrid A. Holm (BCH) |
| **Tentative Senior Author** *(last author)* | TBD |
| **All Other Authors**  | CCHMC – Melanie Myers, Catrina NelsonCHOP – John Connolly, Heather HainColumbia – Hila Milo Rasouly, Julia WynnGeisinger – Alanna Kulchak Rahm; Janet WilliamsHarvard – Kurt Christensen, Emilie ZoltickKPW/UW – David VeenstraMayo – Richard Sharp, Iftikhar Kullo, Joel PacynaNW – Sharon Aufox, Maureen SmithVanderbilt – Ellen Clayton |
| **Sites Participating** | All eMERGE III clinical sites |
| **Background / Significance** | Most eMERGE III sites conducted baseline participant surveys, and all sites conducted post-disclosure surveys of participants receiving positive genomic results to understand the impact of return of genomic information to participants. The Participant survey subgroup has harmonized survey questions across sites to allow for comparing how the impact differs among participants across a variety of health care settings and prior clinical conditions. Responses were collected locally, and the responses to the questions common to all sites were reconciled.Little is known about the psychological impact on patients of receiving positive genomic results when the testing was done in a screening setting. We will assess the psychological impact using the FACToR/MICRA scale and the Decisional Regret scale. |
| **Outline of Project** | We will analyze data from participants with positive results on the 1-month and 6-12 month post-disclosure participant surveys at all sites. Psychological impact of positive results on participants will be assessed using the following outcomes:FACToR/MICRA scaleDecisional regretAnalyses will be conducted to determine if psychological impact (above) is associated with the following predictors:Result related to indication for sequencing or unrelated to known conditions Unselected vs. selected populationPrivacy/confidentiality concerns (HINTS)Decisional conflict at baselineGenderAgeType of disease risk reported (cancer, cardiovascular, other)?Literacy/numeracy/education level Confidence level of the genetic result (Pathogenic, Likely Pathogenic, Risk Factor)Participant site (or by site ROR process – based on the 3 possible processes for ROR found in ROR1 paper)Participant in an integrated healthcare system (Kaiser Permanente Washington, Geisinger, Mayo) compared to those in a university/tertiary care system (BWH, CCHMC, CHOP, Columbia, NW, VUMC)  |
| **Desired Data - Common Variables\*** *(Available from the CC)* | [x] 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)* 1. Answers to the survey questions.
2. The indication for sequencing the participant for the sites that had patients with an indication
3. The gene and condition for the positive result for each participant.
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| **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** | TBD to address the hypotheses |
| **Ethical Considerations** | This survey assesses the risks and benefits of return of uncertain genomic information to participants. |
| **Target Journal** | TBD |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | 1. Formulate data analysis plan – 02/2020
2. Data collection completed from all sites – 02/2020
3. Data analysis completed – 04/2020
4. Draft manuscript – 05/2020
5. Finalize manuscript – 06/2020

Submission to journal – 07/2020 |

**\*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