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| **eMERGE Network: Manuscript Concept Sheet** |
| **Reference Number** *(to be assigned by CC)* | NT426 |
| **Submission Date** | 28 June 2021 |
| **Project Title** | Genetic counselors’ perspectives on returning high-risk PRS results in eMERGE |
| **Tentative Lead Investigator** *(first author)* | Sabrina Suckiel |
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| **All Other Authors**  | Eimear KennyEmily SoperAyuko Iverson\*any eMERGE authors who are interested |
| **Sites Participating** | Open to all eMERGE sites with genetic counselors returning high-risk PRS results |
| **Background / Significance** | The eMERGE IV Network aims to integrate polygenic risk scores (PRS) into clinical care. The majority of the 10 sites in eMERGE IV plan to have genetic counselors (GCs) return high-risk PRS results to patient-participants. However, clinical GCs have had limited exposure to PRS outside of certain cancer conditions. Therefore, little is known about GCs’ experiences with returning PRS results to date. This study will explore the early experiences of GCs in eMERGE with PRS across a variety of common conditions implemented in the study, and across a variety of sites and patient populations. Findings from this study will aid in building best practices in genetic counseling for common disease risk. |
| **Outline of Project** | This manuscript will describe:1. Experiences of GCs with PRS across a variety of conditions in eMERGE
2. Counseling approaches to disclosing PRS results, and how this compares to monogenic result disclosure
3. Challenges in communicating high-risk PRS results
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| **Desired Data - Common Variables\*** *(Available from the CC)* | N/A |
| **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[x] Other (not listed above): **N/A** |
| **Does project pertain to an existing eMERGE Phenotype?** | [ ] Yes, if so please list: [x] No |
| **Planned Statistical Analyses** | A survey that includes both closed- and open-ended questions will be administered to study GC across all sites. Survey data will be reported using descriptive statistics, and themes will be identified in the open-ended survey responses. |
| **Ethical Considerations** | N/A |
| **Target Journal** | HGG Advances (or similar) |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | * August 2021 – manuscript concept sheet approval
* December 2021 – survey development
* April 2022 – survey data collection
* September 2022 – data analysis
* November 2022 – draft manuscript
* December 2022 – submit manuscript
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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): ICD9/10, 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, Blood pressure medications
* Other: Case/Control status on Phase I and Phase II phenotype: only on GWAS dataset participants