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| **eMERGE Network: Manuscript Concept Sheet** | | |
| **Reference Number**  *(to be assigned by CC)* | NT456 | |
| **Submission Date** |  | |
| **Project Title** | **Inclusion and participation of individuals with disability in eMERGE IV** | |
| **Tentative Lead Investigator** *(first author)* | TBD | |
| **Tentative Senior Author**  *(last author)* | Maya Sabatello ([ms4075@cumc.columbia.edu](mailto:ms4075@cumc.columbia.edu)) | |
| **All Other Authors** | Ingrid Holm  Malia Fullerton  Paul Appelbaum  Wendy Chung  Chunhua Weng  Maureen Smith  \*any eMERGE authors who are interested | |
| **Sites Participating** | All eMERGE IV clinical sites | |
| **Background / Significance** | The eMERGE IV prescreening survey includes a question designed to identify potential participants with disabilities, and we are not aware of any other national genomic research consortium that has done so. For individuals who begin but who do not proceed to enrollment, including those who indicated on the prescreening survey that they have a disability, the expectation is that sites will follow up with these individuals to inquire about why they did not enroll, whether this was due to needs for accommodations, and what accommodation may be provided to facilitate enrollment and retention. In addition, the study collected data in the baseline survey about participants’ chronic health conditions such as kidney disease and type 1 diabetes that may be considered as disability under existing US laws and policies. The harmonization of study materials and retention processes across eMERGE sites offers an opportunity to explore the uncharted field of inclusion of people with disabilities in genomic research, including demographic characteristics, health behaviors, and expectations surrounding genetic testing for common diseases. | |
| **Outline of Project** | The participant surveys in eMERGE IV will allow the collection of data across the sites on the questions outlined above, as well as those who have chronic conditions such as kidney disease or type 1 diabetes.  We will analyze survey response data in the prescreen survey from all participants who self-identified as having a disability and those who reported chronic health condition in the baseline survey. We will report those who have been followed up to inquire about continued enrollment, stated needs of accommodation, the provision of reasonable accommodations, and rates of decliners and enrollees among those who have been followed up.  Among those enrolled, we will analyze relevant questions in the baseline and preRoR surveys assessing perceived health risks (baseline survey), health behaviors, experiences with genetic testing and understanding of and expectations from a genetic testing (preRoR survey).  Across analyses, we will compare responses of those who self-identify as having a disability to those who do not and determine if response differences are associated with disability type, comorbidities and/or other relevant intersectionalities (see below).  The specific questions are:  \* Prescreen survey questions on disability, race/ethnicity, gender, participant age, preferred language and interest in participation in eMERGE (separately and comparatively, by demographic characteristics);  \* Responses to questions on accommodation, as recorded by recruitment sites.  \* Baseline survey questions on gender identity, personal health history, knowledge and understanding section, pregnancies & number of live births  \* Pre-RoR survey questions relating to study and other experiences, including those under “genetic testing factors”, “expectation of testing” and SES | |
| **Desired Data - Common Variables\***  *(Available from the CC)* | Demographics  ICD9/10 codes  CPT codes  Phecodes  BMI | Common Variable Labs  Common Variable Meds  Geocoding 2015 ACS variables  Other: Case/Control status |
| **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)*  Answers to the survey questions. | |
| **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** | 1. Descriptive analyses 2. Comparative analyses of those identified as having a disability by disability-type and accommodations, including pre-screening survey responses of those who enrolled vs. those who did not, based on self-reported disability status question and retention based on stated need and provision of reasonable accommodations. 3. Among enrollees with disabilities, compare responses exploring the questions above (“genetic testing factors”, “expectation of testing”) based on disability status and other key demographics, e.g., racial/ ethnic minorities. 4. Other analyses TBD. | |
| **Ethical Considerations** | N/A | |
| **Target Journal** | TBD | |
| **Milestones**  *(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | 1. Manuscript concept sheet approval – 07/2022 2. Formulate data analysis plan – 09/2022 3. Data collection on disability status & preRoR completed at all sites – 03/2023 4. Data analysis – 05/2023 5. Draft manuscript – 06/2023 6. Finalize manuscript – 07/2023   Submission to journal 08/2023 | |

**\*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

***\*\**** *This section should include the timeline for completion of project, including approval, project duration, first and second draft of the paper and submission.*