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| **eMERGE Network: Manuscript Concept Sheet** | | |
| **Reference Number**  *(to be assigned by CC)* | NT277.1 | |
| **Submission Date** | 4/16/2019 | |
| **Project Title** | Operationalizing participant choices about genomic results: Beyond all or none ACMG recommended genes | |
| **Tentative Lead Investigator** *(first author)* | L. Rasmussen | |
| **Tentative Senior Author**  *(last author)* | K. Marsolo | |
| **All Other Authors** | Investigators at sites involved in operationalizing participant choices – M.F. Myers, M.E. Smith, W. Chung, H. Hakonarson, S.A. Aufox, J. Wynn, J. Connolly; investigators at central laboratories – H. Zouk, E. Venner, Y. Yang, D. R. Murdock, Y. Jiang | |
| **Sites Participating** | CCHMC, Northwestern, Columbia, CHOP, LMM, Baylor, other sites where participants were offered choices about secondary results (all or none vs. some of the ACMG 56/59 genes) | |
| **Background / Significance** | In clinical practice, patients are typically given the option to learn all or zero secondary sequencing results recommended by the ACMG. As the field becomes more comfortable with the return of sequencing results, and we move towards more patient-centered care, it is necessary to provide patients with more granular choices in deciding what to learn about their panel results. There is considerable variation between sites regarding study participants’ engagement in prospectively choosing the type of e3 sequencing panel results they want to learn. Investigators at several eMERGE 3 sites were challenged to create tools that enabled participants’ granular choices. | |
| **Outline of Project** | 1. Describe and compare how site-specific returnable genes were categorized for participant choices    1. Rationale for allowing granular choices    2. Compare proportions of participants across sites who made granular choices    3. Successes, challenges and limitations of categorizing 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  **NONE** |
| **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)*  Selections made by participants; optionally, any developed resources (consent forms, sample reports, etc.) that illustrate the collection and operationalization of choices | |
| **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):  **NONE** | |
| **Does project pertain to an existing eMERGE Phenotype?** | Yes, if so please list  No | |
| **Planned Statistical Analyses** | Content analysis and descriptive statistics | |
| **Ethical Considerations** | None | |
| **Target Journal** | GIM or JAMIA | |
| **Milestones**  *(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | Approval – Spring 2018  Data collection – early Summer 2018  First draft – Fall 2018  Submit for publication – Winter 2018 | |

**\*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