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| **eMERGE Network: Manuscript Concept Sheet** | | |
| **Reference Number**  *(to be assigned by CC)* | NT300 | |
| **Submission Date** | 07/25/2018 | |
| **Project Title** | Understanding the return of results process: Content review of patient summary letters | |
| **Tentative Lead Investigator** *(first author)* | CCHMC: John Lynch, PhD | |
| **Tentative Senior Author**  *(last author)* | Geisinger: Janet L. Williams, MS, LCGC | |
| **All Other Authors** | Harvard: Carrie Blout, MS, LCGC;  Geisinger: Alanna Kulchak Rahm, PhD, MS, LCGC; Amy Sturm, Md, LCGC; Adam Buchanan, MPH, MS, LCGC; Marc S. Williams, MD; Tara Schmidlen, MS, LCGC; Cassandra Pisieczko;  Boston Children’s Hospital: Ingrid Holm, MD; Jill Madden, MS;  Northwestern: Maureen Smith, MS, LCGC; Sharon Aufox, MS, LCGC; Christin Hoell, MS, CGC;  Columbia: Julia Wynn, MS, LCGC; Hila Milo Rasouly, PhD, MS, CGC; Maddalena ?  CCHMC: Cindy Prows, RN; Melanie Myers, PhD, MS, LCGC  Meharry: Rajbir Singh, MD;  Vanderbilt: Georgia Wiesner, MD  Kaiser WA: Gail Jarvik, MD; Kathy Leppig, MD;  Mayo: David Kochan;  Marshfield: Scott Hebbring, PhD | |
| **Sites Participating** | Open to all sites. Those who have responded:  BCH  Columbia  CCHMC  Geisinger  Harvard  Kaiser WA  Mayo  Marshfield  Meharry  Northwestern  Vanderbilt | |
| **Background / Significance** | eMERGE 3 is charged with the development of processes to communicate research genomic results to patients and providers with placement of sequence results in the EHR. Often this information is unexpected when there was no prior indication for the sequencing. One important vehicle for patient communication is the results summary letter. Each eMERGE site has autonomy to build the process that best fits their health system environment and has independently formulated patient result letters. It is likely that the letters and/or web content varies not only by site but also based on the gene, variant, and condition identified. In addition, a few sites are returning negative sequence results. Several eMERGE investigators have previously evaluated patient and provider preferences for communication of laboratory results. We propose to review the current communication materials used by programs to communicate eMERGE genomic sequencing results for common elements, language choices, literacy level, concepts addressed, and resources provided. Through this process we will develop recommendations for the communication of results to patients receiving genomic information. Evaluation of the various elements of the site-specific processes will contribute to the body of knowledge necessary to formulate evidence-based practice guidance for communicating genomic sequencing results. | |
| **Outline of Project** | We will seek a representative from each site to supply the documents and or links to information provided to their participants who receive results. The information to review may include summary letters of positive, negative and VUS results as well as condition specific information and letters to share with family members regarding risk and testing options. The information will be evaluated by qualitative content analysis, literacy assessment, and compilation of resources will be reviewed. We will use this information to make recommendations regarding what information to be included with the return of results to patients undergoing genomic sequencing. | |
| **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)*** | Common elements or sections  Literacy level  Resources provided  Method to deliver communication  Inventory of patient information materials at each site  Common themes  Common language or phrases | |
| **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** | NA/Descriptive Study | |
| **Ethical Considerations** | None | |
| **Target Journal** | GIM or Journal of Genetic Counseling | |
| **Milestones**  *(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | Gather materials by 9/01/2018  Content assessment by 12/31/2018  Manuscript submitted by 1/31/2019 | |

**\*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