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| **eMERGE Network: Manuscript Concept Sheet** |
| **Reference Number** *(to be assigned by CC)* | NT352 |
| **Submission Date** | June 20, 2019 (revised January 24, 2020) |
| **Project Title** | A case series of lessons from eMERGE on genomic implementation |
| **Tentative Lead Investigator** *(first author)* | Casey Overby Taylor |
| **Tentative Senior Author** *(last author)* | Sandy Aronson |
| **All Other Authors**  | Lead authors of case report proposal submissions: BWH/Matt Lebo, NU/Luke Rasmussen or Laura Rasmussen-Torvik, CU/Chunhua Weng and David Fasel, JHU/Nara Sobreira and Casey Overby Taylor, MC/Hana Bangash and Iftikhar Kullo, BCM/Mullai Murugan and Eric Venner, CCHMC/Cindy Prows and Eric Hall  |
| **Sites Participating** | We propose a series of case studies to illustrate experiences of institutions implementing genomic medicine programs with using clinical information systems to return results from genomic testing to patients and providers.  |
| **Background / Significance** | Case report submissions describe what was done, Iessons learned, and implications of those efforts. The reports can illustrate, explore, report, analyze, summarize, challenge, or describe practical work carried out to address the problem of returning genomic test results. Topics of interest to clinical informatics and health IT communities such as usability, workflow efficiency, optimizing implementation, governance, informatics infrastructure, interoperability, data privacy and security, etc should be explored. |
| **Outline of Project** | **Format suggestion for submission*** Title (20 word limit
* Abstract (300 word limit)
* Author Keywords (10 max)
* Method: What we did
* Findings: What we learned
* Discussion: What it means, how others might use the findings, implications for practice
* Conclusion & What’s next
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| **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)*** | *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[ ] Other (not listed above): |
| **Does project pertain to an existing eMERGE Phenotype?** | [ ] Yes, if so please list [x] No |
| **Planned Statistical Analyses** | None (A content analysis of EHRI WG meeting minutes and milestone tracker records) |
| **Ethical Considerations** | None |
| **Target Journal** | ACI Open |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | Prepare rough drafts – Feb 28th, 2020Submit to ACI Open for review – end of March 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