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| **eMERGE Network: Manuscript Concept Sheet** |
| **Reference Number***(to be assigned by CC)* |  NT380 |
| **Submission Date** | March 10th, 2020 |
| **Project Title** | Genomic Considerations for FHIR: Lessons from the eMERGE Implementation |
| **Tentative Lead Investigator***(first author)* | Mullai Murugan |
| **Tentative Senior Author***(last author)* | Richard Gibbs |
| **All Other Authors** | Larry Babb, Luke Rasmussen, Casey Overby Taylor, Eric Venner, Fei Yan, Victoria Yi |
| **Sites Participating** | Baylor College of Medicine, Broad Institute, Northwestern University, Johns Hopkins University |
| **Background / Significance** | The need for structured representation of the eMERGE clinical genetic results was a necessary first step to achieve the eMERGE network’s phase III milestone of EHR integration and furtherance of clinical decision support. The network opted to use the readily available GeneInsight XML format for standardized and structured representation of genetic results. However, with an eye to the future and committed to growing a national standard, as part of its year five goals, the network formulated an objective to evaluate the use of HL7 FHIR as a standard to represent clinical genomics results particularly considering the increasing expediency of HL7 FHIR as an international healthcare interoperability standard. This manuscript will illustrate our efforts towards the creation of a FHIR based genomics specification for eMERGE and the implementation of a proof of concept EHR integration pilot to illustrate the feasibility of the specification and its capability to aid with clinical decision support. Additionally, this effort will also detail our collaboration with the HL7 Clinical Genomics Workgroup, our use of of the Genomics Reporting Implementation Guide for the eMERGE FHIR specification, lessons learned, the diversity of the FHIR technology landscape, associated implementationchallenges and our recommendations for the future. |
| **Outline of Project** | 1. Background on the current state of standards for clinical genomic reporting
2. Expediency of a HL7 FHIR based genomics standard for eMERGE
3. Design of a HL7 Clinical Genomics Reporting Implementation Guide based FHIR specification for eMERGE
4. EHR integration and Clinical Decision Support with an implementation pilot
5. Lesson learned, feasibility/validity analysis and future considerations
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| **Desired Data - Common Variables\****(Available from the CC)* | * Demographics ☐Common Variable Labs
* ICD9/10 codes ☐Common Variable Meds
* CPT codes ☐Other: Case/Control status on Phase I
* Phecodes and Phase II phenotypes
* BMI
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| **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)*None |
| **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):~190 Northwestern University’s eMERGE III Clinical Reports used as source data setN/A |
| **Does project pertain to an existing eMERGE Phenotype?** | * Yes, if so please list

☒No |
| **Planned Statistical Analyses** | None, this paper primarily focuses on creating a HL7 FHIR based standard specification for eMERGE clinical reporting. |
| **Ethical Considerations** | None |
| **Target Journal** | JAMIA |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | Initial Draft: April 2020Revisions by Co-Authors: April 2020Final Draft reviewed by Co-Authors: May 2020 Final Draft Submitted: May 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