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| **eMERGE Network: External Collaborator Manuscript Concept Sheet** | | |
| **Reference Number**  *(to be assigned by CC)* | NT319 | |
| **Submission Date** | 12/18/2018 | |
| **Project Title** | Use of Infobuttons to Find Answers to Clinician’s Questions in Clinical Genomics | |
| **Tentative Lead Investigator** *(first author)* | Michael Watkins | |
| **Tentative Senior Author**  *(last author)* | Guilherme del Fiol | |
| **eMERGE Site Sponsor & Contact** | Luke Rasmussen | |
| **All Other Authors** | Casey Overby Taylor, Marc Williams, Bob Freimuth, Nephi Walton, John Connolly, Darren Johnson, Bret Heale | |
| **Sites Participating** | University of Utah, Intermountain Healthcare, Northwestern University, Geisinger, Johns Hopkins, Mayo, CHOP | |
| **Background / Significance** | Existing work within eMERGE regarding infobuttons (context-sensitive links embedded in an EHR or patient portal) has resulted in recommendations for how genomic knowledge resources should be structured (Overby et al., 2014). In addition, the development of the DocUBuild system has provided a technical platform which can aid knowledge resource authors in creating content that is more optimally retrieved using infobuttons. Previous works have also elicited clinicians’ questions in clinical genomics (Heale et al) and investigated the readiness of genomic knowledge resources to be integrated with EHR systems through the HL7 infobutton standard (Heale et al). A current gap is to identify technical approaches to retrieve relevant content from genomic resources that answers clinical genomic questions within the EHR as well as content gaps in those resources. The study findings will allow us to identify systematic approaches to deliver high priority clinical genomics knowledge into EHRs and help prioritize content development in DocUBuild, for questions that are difficult to answer or not answered in available clinical genomic resources. | |
| **Outline of Project** | Identify a set of common clinicaI questions requiring genomic knowledge resources.   * Start with published literature, then collectively review and complete the set.   Identify genomic knowledge resources which help answer each question.   * Include a mix of resources such as PharmGKB, GeneReviews, and UpToDate.   For each question, identify specific content within each resource that provides an answer to the question.   * Annotate articles with context, where present. Focus is on finding the most appropriate level of detail to answer each question. * All annotations will be done by 2 reviewers independently. Disagreements will be resolved through consensus with the DocUBuild team.   Analyze the ability of an Infobutton request to retrieve the answers identified above for each resource.  Analyze and summarize results.  Prepare and submit manuscript. | |
| **Desired Data - Common Variables\***  *(Available from the CC)* | Demographics  ICD9/10 codes  CPT codes  Phecodes  BMI  **None** | 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)*  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):  **None** | |
| **Does project pertain to an existing eMERGE Phenotype?** | Yes, if so please list  No | |
| **Planned Statistical Analyses** | Summary statistics on prevalence of context elements | |
| **Ethical Considerations** | The study will only use freely available resources, in accordance with respective usage policies outlined at each resource. | |
| **Available Funding or Resources** | Michael Watkins and Dr. Guilherme Del Fiol’s time is covered under existing projects and academic activities. | |
| **Target Journal** | Applied Clinical Informatics | |
| **Milestones**  *(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | January 2019 – Begin annotation work  March 2019 – Data analysis  May 2019 – Prepare first draft of manuscript  July 2019 – First draft review from co-authors  August 2019 – Revise manuscript  September 2019 – Final review / feedback from authors  October 1, 2019 – Submit manuscript | |

**\*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