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| **eMERGE Network: Proposal for Analysis**Project/Manuscript Concept Sheet |
| **Reference Number** | NT296 |
| **Submission Date** | June 26th, 2018 |
| **Project Title** | Collection and Analysis of Large-Scale Outcome Measures following Targeted Next Generation Sequencing |
| **Tentative Lead Investigator** *(first author)* | Josh Peterson |
| **Tentative Senior Author** *(last author)* | Marc WilliamsHakon Hakonarson |
| **All Other Authors**  | Outcome leads from each eMERGE implementation site are expected to participateOther eMERGE investigators with interest in Outcomes.Jyoti Dayal (NGHRI), Cindy Prows (CCHMC), Alanna Rahm (Geisinger), Georgia Wiesner (VUMC), Janet Williams (Geisinger),  |
| **Sites Involved** | All eMERGEseq Implementation sites |
| **Background / Significance** | Methods to collect Outcomes across the broad array of disease risk The eMERGE Outcomes workgroup was formed to track cross-site outcomes following return of disease risk variants from the targeted sequencing platform, eMERGEseq. The workgroup focused on answering whether eMERGEseq impacted health care utilization, intermediate biomarker indicators of future disease effect, and clinical outcomes of importance to patients and families. Over the course of 18 months, the workgroup developed 18 data collection instruments containing 1410 fields. The instruments covered return of results, family cascade testing, and specific outcome forms tailored to variant sets grouped according to the area of disease. Instruments were designed to collect information 6 months following Return of Results as represented at each site’s Electronic Health Record (EHR). The manuscript will review the process of developing the instrument questions, the content, and discuss the potential for scaling the process to even larger cohorts. |
| **Outline of Project** | * Conceptual approach / model
* Outcome measurement strategy
* Infrastructure
* Outcome development process
* Return of results and familial cascade testing forms
* Common elements of each disease specific form
* Protocol for EHR abstraction
* Discussion of future approaches with greater automation
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| **Desired Variables** *(essential for analysis**indicated by* ***\*****)* | Demographics of eMERGE cohort |
| **Desired Data** | Outcome data collection formsExpected recordsPreliminary return rates |
| **Planned Statistical Analyses** | Descriptive summaries of expected scale of return of resultsStatistical approaches to analyses based on genotype and phenotype categories to be discussed.Heterogenity of studies aggregated for outcomes to be addressed. |
| **Ethical Considerations** | None – no patient level data to be collected for this protocol paper |
| **Target Journal** | 1. Genetics in Medicine (reach)
2. eGEMS
3. Journal of Medical Genetics
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| **Milestones\*\*** | Approval: July 15th, 2018Duration: 6 monthsFirst draft: September 30th, 2018Second draft: October 30th, 2018Submission: December 1st, 2018 |

***\*\**** *This section should include the timeline for completion of project, including: approval, project duration, first and second draft of the paper and submission.*