**eMERGE Network Proposal for Analysis**

Project/Manuscript Concept Sheet

|  |  |
| --- | --- |
| **Reference Number** | NT271 |
| **Submission Date** | January 30, 2018 |
| **Project Title** | Mayo Clinic Genomic Medicine Implementation Study: Design and Initial Results |
| **Tentative Lead Investigator (first author)** | Iftikhar Kullo |
| **Tentative Senior Author (last author)** | Stephen Thibodeau (Mayo Clinic) |
| **All other authors**  | Mayo team and Baylor team |
| **Sites Involved** | Mayo Clinic and Baylor College of Medicine |
| **Background / Significance** | We describe design and initial results of Mayo’s genomic medicine project conducted as part of eMERGE III: The Return of Actionable Variants Empiric (RAVE) study. |
| **Outline of Project** | We recruited 3030 participants from Mayo Clinic biobanks in Rochester, MN, who had hypercholesterolemia, colon polyps or both, thereby enriching for FH and monogenic causes of CRC. DNA of 2538 samples was sent to Baylor College of Medicine Human Genome Sequencing Center (BCM-HGSC), a Central Laboratory Improvement Amendment (CLIA)-certified facility, for targeted sequencing using the eMERGEseq panel. **Once sequencing data (for research) for Mayo participants** were available, variant identification was completed and actionable variants in cardiovascular genes were identified using an in-house bioinformatics pipelines. We will perform a preliminary analyses of whether expected phenotypes are present in participants with P/LP variants in CV disease genes. (Note: We will not be using Clinical Research Reports from BCM for this preliminary analysis; also ONLY Mayo data are being used)  |
| **Desired****Variables (essential for analysis****indicated by \*)** | Additional Variables:Sequence data (eMERGEseq) for 2537 Mayo Rochester participants (already downloaded)EMR review of Mayo participants |
| **Desired data** |  |
| **Planned Statistical Analyses** | NA |
| **Ethical considerations** | None noted |
| **Target Journal** | Mayo Clinic Proceedings |
| **Milestones\*\*** | November-December 2017: Data summaryJanuary 2018: Manuscript preparationFebruary 2018: Manuscript submission |

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