**eMERGE Network Proposal for Analysis**

Project/Manuscript Concept Sheet

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| **Reference Number** | NT242 |
| **Submission Date** | 6/28/17 |
| **Project Title** | Clinical outcomes after screening for cardiomyopathy genes |
| **Tentative Lead Investigator (first author)** | Christin Hoell |
| **Tentative Senior Author (last author)** | Laura Rasmussen-Torvik |
| **All other authors** | Jennifer Pacheco, Lisa Castillo, Dr. Beth McNally, Maureen Smith, any eMERGE investigator interested in the project |
| **Sites Involved** | All eMERGE sites |
| **Background / Significance** | There are several genes on the eMERGE3 platform that have rare genetic variants known to be associated with cardiomyopathies (see below). We seek to examine how clinical care changes after identification of rare variants potentially associated with cardiomyopathies. |
| **Outline of Project** | 1. Identify individuals with known pathogenic and likely pathogenic variants genes listed below at all participating sites 2. Extract information on clinical care pre and 6 months post result return as detailed in the cardiomyopathy outcomes collection tool and chart extraction list below 3. Synthesize information within and between sites |
| **Desired**  **Variables (essential for analysis**  **indicated by \*)** | All variables on the cardiomyopathy outcomes collection tool as well as variables collected by chart extraction listed below. |
| **Desired data** | Emerge 3 .vcfs, information about clinical variants returned at each of the sites. Site RedCap data from cardiomyopathy outcomes collection tool as well as chart extraction data. |
| **Planned Statistical Analyses** | Paper will generally rely on descriptive data, presenting counts in a pre-post format. If power allows, paired t-tests or chi squares may be used to assessed statistical significance. |
| **Ethical considerations** | Care must be taken in the abstraction of clinical records to maintain patient confidentiality. |
| **Target Journal** | TBD |
| **Milestones\*\*** | January 2019 – data collection complete  February 2019 – data analysis complete  March 2019 - first draft complete  May 2019 – submit for publication |

ACMG cardiomyopathy genes: *MYBPC3*

*MYH7*

*TNNT2*

*TNNI3*

*TPM1*

*MYL3*

*ACTC1*

*PRKAG2*

*MYL2*

*LMNA*

*GLA*

*PKP2*

*DSP*

*DSC2*

*TMEM43*

*DSG2*

Chart Extraction:

1. All computed variables from all EKG
2. The following measures from all Echocardiograms:
   1. EF
   2. LVIDs
   3. LVIDd
   4. LV wall thickness
   5. Septal wall thickness
   6. Sinus of Valsalva diameter
3. Prescriptions for Beta Blockers, Ca Channel blockers, ACE inhibitors, ARBs
4. Procedure diagnosis codes for the following:
   1. Afib
   2. Heart failure
   3. Stroke
   4. Sudden death
   5. Aborted cardiac arrest
   6. Arrhythmia
   7. Heart transplant
   8. Defibrillator implant
   9. Pacemaker implant
   10. Cardiac ablation
   11. Cardiac cardioversion