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| **eMERGE Network: Manuscript Concept Sheet** |
| **Reference Number** *(to be assigned by CC)* | NT420 |
| **Submission Date** | 3/18/2021 |
| **Project Title** | Probing the polygenic architecture of left atrial diameter |
| **Tentative Lead Investigator** *(first author)* | Ali Manouchehri |
| **Tentative Lead Investigator Email Address** | ali.m.manouchehri@vumc.org  |
| **Tentative Senior Author** *(last author)* | Jonathan Mosley |
| **All Other Authors**  | Quinn Wells, RJ Vaitinadin, Mingjian Shi, Vineet Aggarwal, Jane Ferguson |
| **Sites Participating** | Current participants: Vanderbilt Open to all sites |
| **Background / Significance** | The left atrium (LA) plays a major role in cardiac physiology by collecting blood during systole and modulating left ventricular filling during diastole and preserves pulmonary capillary circulation from hemodynamic overload. A main determinant of left atrial diameter and volume is ventricular diastolic function. The left atrium is exposed directly to left ventricular diastolic pressure and dilates with increasing pressure. The degree of left atrial enlargement is associated with adverse prognosis in different clinical settings.  |
| **Outline of Project** | To identify clinical phenotypes associated with a polygenic score for LA diameter developed in a VUMC clinical population.1. Perform a PheWAS using a polygenic score developed from a population of ~16,000 European Ancestry individuals who has LA diameter measured by TTE as part of routine clinical care at VUMC.
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| **Desired Data - Common Variables\*** *(Available from the CC)* | [x] Demographics [x] ICD9/10 codes[ ] CPT codes[ ] Phecodes[ ] BMI | [ ] Common Variable Labs[ ] Common Variable Meds[ ] Other: Case/Control status on Phase I and [ ] Phase II phenotypes |
| **Other Desired Data *(Available from participating sites)*** |  |
| **Desired Genetic Data** | [x] 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): |
| **Does project pertain to an existing eMERGE Phenotype?** | [ ] Yes, if so please list [x] No |
| **Planned Statistical Analyses** | PheWAS |
| **Ethical Considerations** | None |
| **Target Journal** | Will be determined by the results, but anticipate an AHA journal. |
| **Milestones***(This section should include the key dates for completion of project, including approval, project duration, draft completion, and submission.)* | Gather data from coordinating center: 04/01/2021Conduct statistical analyses: 04/5/2021Write manuscript: 04/15/2021Circulate and submit manuscript: 04/30/2021  |