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| **eMERGE Network: Manuscript Concept Sheet** | | |
| **Reference Number**  *(to be assigned by CC)* | NT361 | |
| **Submission Date** | 09/25/2019 | |
| **Project Title** | Genome-wide association of Nephrotic Syndrome | |
| **Tentative Lead Investigator** *(first author)* | Jacklyn N. Hellwege | |
| **Tentative Senior Author**  *(last author)* | Katalin Susztak (U Penn) and Todd Edwards (Vanderbilt) | |
| **All Other Authors** | Digna Velez Edwards, Josh Denny, Dan Roden | |
| **Sites Participating** | Open to all sites  Current participants: Vanderbilt | |
| **Background / Significance** | Nephrotic syndrome is a collection of disorders wherein the kidneys allow too much protein in the urine, combined with hypoalbuminuria and often hyperlipidemia and edema. This disease impacts 16 per 100,000 individuals, with idiopathic nephrotic syndrome being the most common form among youth. To date, the largest GWAS that has been performed includes only 450 cases. | |
| **Outline of Project** | We propose to utilize eMERGE and BioVU data to perform the largest GWAS of Nephrotic Syndrome to date (>1500 cases). We will use ICD 9/10 codes and phecodes to describe cases and controls, respectively. We plan to perform analyses adjusted for covariates and stratified by race, as well as performing follow-up PheWAS analyses. | |
| **Desired Data - Common Variables\***  *(Available from the CC)* | Demographics  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** | 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  No | |
| **Planned Statistical Analyses** | GWAS, PheWAS | |
| **Ethical Considerations** | None | |
| **Target Journal** | Depends on results, possibly a genetics journal such as Human Molecular Genetics or a general journal such as elife, Nature Communications, or Scientific Reports, a renal journal such as JASN. | |
| **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: 10/2019  Conduct statistical analyses: 11/2019  Write manuscript: 1-3/2020  Circulate and submit manuscript: 4/2020 | |