Clinical Utility of Prenatal Whole Exome Sequencing
NCT03482141
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Statistical analysis plan: Our study design will allow statistical comparisons that will generate useful data regarding WES implementation. Diagnostic yield will be calculated per case for the proband. It will be based on the number of cases with a positive or likely positive exome result divided by the total number of cases. The size of the "positive" (likely positive or probable positive), "inconclusive" or "negative" groups will depend on the diagnostic success rate of WES in this prenatal cohort. We will conduct analysis to determine diagnostic yield, which may vary based on a number of factors, including setting (prenatal or pediatric). We will tabulate results by disease indication, by race/ethnicity, and by socioeconomic status. Such stratified analyses will be descriptive, and will depend on the actual observed sample sizes within each stratum. Because this is a descriptive study in the prenatal population, we have not developed any hypothesis to test statistically.

Please note that this is the second clinical trial registered for pregnant individuals (prenatal population), funded by the same NIH grant. We submitted results for the other protocol, NCT03525431 for pediatric patients.