

Clinical Utility of Pediatric Whole Exome Sequencing  
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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 our cohort. We will conduct analysis to determine how clinical utility (measured as diagnostic yield) may vary based on a number of factors, including setting (prenatal or pediatric), by disease indication, by race/ethnicity, and by socioeconomic status. Such stratified analyses will depend on the actual observed sample sizes within each stratum.