(virtual) testing will facilitate research during the covid-19 pandemic and are especially well-suited for data collection in rare disease populations.

Translational Science, Policy, & Health Outcomes Science

11979

Using whole-exome and mtDNA sequencing to develop a testing algorithm for diagnosis of mitochondrial disease in Puerto Ricans

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ABSTRACT IMPACT: Alterations in mitochondrial metabolism affect any tissue, especially those with the highest demand for energy. As the symptoms and clinical manifestations are heterogenous, disease diagnosis is challenging. The implementation of genetic-first approach in the diagnosis of mitochondrial diseases will expedite confirmation, treatment, management, and counseling of affected Puerto Rican individuals. OBJECTIVES/GOALS: Mitochondrial diseases are rare, and diagnosis is complex due to the heterogeneity of clinical manifestations. We aim to develop and implement a testing algorithm using a genetics-first approach, facilitating the identification of variants that contribute to mitochondrial disease's etiology and influence onset and progression in Puerto Ricans. METHODS/STUDY POPULATION: This is a cross-sectional study for characterizing clinical laboratory results from profiles used to evaluate metabolic diseases in individuals with suspected mitochondrial disorders from 2018 to 2021. A subset of 25 individuals from biochemical profile will be recruited to analyze their medical and family history, metabolic biomarkers in blood and urine, hearing test, imaging and chromosomal microarray. The implementation of a genetic testing algorithm using whole exome and mitochondrial DNA sequencing will be performed in a subset of 11 randomized individuals. Descriptive analysis will be reported, including a catalog of all variants. Multivariate analysis will be performed to estimate the statistical association between variants and phenotypes reported and adjusting for potential confounders. RESULTS/ANTICIPATED RESULTS: The biochemical profile of pediatric Puerto Rican individuals suspected of having mitochondrial diseases will be altered and can be used to differentiate among other metabolic causes. We expect to find altered levels of lactate, pyruvate and carnitines in serum, as well as altered organic acids in urine. The implementation of a testing algorithm using both, mitochondrial DNA and whole exome sequencing as first approach will be enabling the identification of disease-causing variants, thus enhancing and confirming the diagnosis of mitochondrial disease in Puerto Ricans. We will be able to identify rare/novel variants specific to our Hispanic population, for both nuclear and mitochondrial DNA. DISCUSSION/SIGNIFICANCE OF FINDINGS: This study will help to characterize the metabolic profile of pediatric Puerto Ricans. No previous study has been reported that describes testing algorithms for genetic diagnosis of mitochondrial disease in our population. Variants found will contribute to a deep understanding of the genetic contribution to phenotypes and disease susceptibility.

92811

Implementation of the Fitness, Lifestyle, and Optimal Wellness (FLOW) Program and Its Associated Health Outcomes

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ABSTRACT IMPACT: Through its interdisciplinary, tailored approach, the FLOW program could change the way that we approach promoting healthy lifestyle changes in the primary care field. OBJECTIVES/GOALS: The goal of this project is to assess patient outcomes associated with the implementation of the Fitness, Lifestyle, and Optimal Wellness (FLOW) Program. The ultimate aim of this program is two-fold: increasing patient-reported wellness and improving objective health measurements. METHODS/STUDY POPULATION: The FLOW program consists of a multidisciplinary team of sports medicine physicians, nutritionists, fitness trainers, and clinical psychologists. Patients who choose to participate in the program undergo a comprehensive physician-guided assessment, including lifestyle and metabolic evaluation, biomarker profile, and body composition analysis. Based on the patient's goals and results of evaluation, he/she is then connected with other members of the FLOW team to develop a comprehensive plan and offer resources for potential improvements in physical activity, nutrition, and/or behavior. The patient will undergo follow-up assessments and questionnaires at three and six months to track their objective measurements and reported progress. RESULTS/ANTICIPATED RESULTS: The anticipated results of the FLOW program are an overall improvement in patient health and wellbeing. More specifically, we anticipate seeing increased levels of exercise from initial reported levels, as well as better nutrition habits. We expect to see improvements in follow-up body composition assessments, with gains in fat-free mass and decreased body fat, in addition to patient-reported improvements in behavioral health as measured by PHQ-9, GAD-2, and the Perceived Stress Scale. We will also assess reported sleep health with the hopes to see improvement in follow-up assessments. DISCUSSION/SIGNIFICANCE OF FINDINGS: The FLOW program is designed to address health inequities that disproportionately affect the Deep South. Through this program, we propose a new role of the primary care team in promoting healthy lifestyle habits and disease prevention through exercise, nutrition, and behavioral health services.
Regulatory Science

Regulatory Science

Clinical Trial

95347

Examining the Impact of the BPCA: Promoting Pediatric Inclusion in Clinical Trials and Pediatric-Specific Drug Information

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ABSTRACT IMPACT: It provides insight in the relationship between pediatrics and clinical research and how pediatric

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