



Characterization of Distal Myopathy and Dysphagia in Nephropathic Cystinosis and Evaluation for Muscle Regenerative Capacity Research Study

About the Study: Dr. Reza Seyedsadjadi and Dr. Florian Eichler of the Massachusetts General Hospital (MGH) Center for Rare Neurological Diseases are conducting a study to learn more about muscle weakness and swallowing difficulties in Nephropathic Cystinosis and the ability of these muscles to regenerate (rebuild themselves). This study is currently funded by the Cystinosis Research Foundation (CRF).

There are three primary goals of this study:

1. To better understand how Nephropathic Cystinosis affects muscles used in speech and swallowing. Tests that are currently used to assess swallowing difficulty, such as the Penetration-Aspiration Scale, are not sensitive enough to detect changes over time. Therefore, will use more modern and sensitive techniques to assess how swallowing is affected.
2. To better understand how Cystinosis affects muscles of the arms and legs by using electromyography (EMG), which is a test to measure muscle activity. We hope to determine which muscles are the weakest in Cystinosis patients, as these muscles will be targets for potential future stem/satellite cell technology.
3. To characterize the disease-causing features of skeletal muscle in Cystinosis patients and the ability of this muscle to regenerate. Skeletal muscle has the remarkable ability to regenerate throughout lifetime, due to stem cells called the satellite cells, which activate when skeletal muscle is damaged. In this study, we will collect muscle samples from patients with Nephropathic Cystinosis and provide the tissue to the Rubin Lab at the Harvard Stem Cell Institute, where the tissue will be used to understand what features cause muscle weakness in Cystinosis patients.

What the Study Involves: Participation in this study would involve one visit to MGH. This visit would include the following assessments: a brief neurological examination, an assessment of speech and swallowing, completion of surveys, EMG, and an optional needle muscle biopsy. Patients will receive counseling for their results and the results will be available to his/her care team. Referrals will also be made to a treating physician if further management of nutrition and hydration are necessary. If necessary, patients will also be referred for ongoing swallowing treatment. If you are traveling to Boston to participate, you will be reimbursed up to \$500 for travel and hotel expenses for each study visit. Other travel discounts and funding may be available.

Benefits of Participating: Participation in this study will increase our knowledge of muscle weakness and swallowing difficulty in Nephropathic Cystinosis. Determining the most affected muscles and the ability of skeletal muscle of patients with Cystinosis to regenerate will inform the ability to conduct a future exploratory cell therapy study aimed to treat distal muscle weakness in cystinosis patients. You will not be involved in treatment at this phase, however we hope that the results from this research will lead to the development of future treatment trials.

Who May Participate: You are eligible to participate if you are an adult 18-70 years of age with a diagnosis of nephropathic cystinosis and distal myopathy.

If you would like to participate in this study, or if you would like more information, please contact research coordinator Natalie Grant at 617-643-3799 or nrgrant@mgh.harvard.edu.