

Cystinosis Research Foundation

Lay Abstract Template for Awardees

Please complete this lay-oriented grant abstract form which will be published on the CRF web site, in CRF Star Facts and in the CRF magazine when we announce your grant award. *Please do not exceed 400 words (no more than 1-1/4 page total).* Please submit this form electronically to nstack@cystinosisresearch.org as a Word document.

Principal Investigator (s): Reza Seyedsadjadi

Project Title: Further Characterization of Nephropathic Cystinosis

Objective/Rationale: Please write a lay-oriented statement of the scientific rationale for this project. Approximately 75-85 words.

Myopathy and dysphagia are common symptoms of nephropathic cystinosis. Preceding research has been done to assess which swallowing phases are most affected in patients experiencing these manifestations. The past phases of this project have shown that patients had the most improvement with bolus transport, timing of their pharyngeal swallowing response, and laryngeal elevation with respiratory training and exposure to their physiology. These improvements may be related to the focused effort from the patients who received the respiratory treatment and education on swallowing mechanics.

Project Description: Please write a brief, lay-oriented description of how you will carry out the project. Approximately 125-135 words.

Patients will come on site to Massachusetts General Hospital (MGH) to meet with a neurologist and speech pathologist who will perform baseline procedures such as a neurological exam, muscular ultrasound, and MBSImp. After the conclusion of these procedures we will randomized the patients into three cohorts, each with a different treatment targeting different muscular groups. The patients will be educated on their treatment method and how to progress their training in-between visits. They will report progress with the study coordinator every three months and receive any necessary feedback on their exercise program. The patients will return to MGH one year after their baseline visit to complete the same procedures. The study team will compare the data from both visits across all three treatment arms to assess improvements in function and quality of life.

Relevance to the Understanding and/or Treatment of Cystinosis: Please explain how the project will impact cystinosis treatment or increase our understanding of cystinosis. Approximately 75-80 words.

The imaging methods – MBSImp and muscular ultrasound - will be used to identify and compare which muscles are most affected by myopathy and dysphagia in this patient population. In the interim between on-site visits, the patients are in three separate cohorts using treatments that strengthen three different groups of muscles involved in swallowing. After they complete the second visit, we will be able to increase our understanding of what kind of treatments work best to maintain or increase quality of life.

Anticipated Outcome: Please write a lay-oriented description of what you expect to learn/discover. Approximately 75-80 words.

Prior research has shown that when patients are exposed to their physiological data and make educated, focused efforts to improve function, it may influence their ability to swallow. Given that these patients are practicing their treatment over the course of the year, we expect them all to improve their swallowing function as their musculature is strengthened. We are hoping to discover differences in improvements between the groups with data showing one treatment arm had better/more improvement between baseline and the last on-site visit.