MASSACHUSETTS GENERAL HOSPITAL
NEUROLOGY

HARVARD MEDICAL SCHOOL

To Whom it May Concern,

We are writing to let you know about a research study that might interest you. The Massachusetts General Hospital (MGH) Neuromuscular Clinic is looking for participants for an interventional study to further characterize the mechanism of muscular involvement in swallowing in patients with nephropathic cystinosis experiencing myopathy.

Nephropathic cystinosis is a rare autosomal recessive lysosomal storage disorder due to mutations in the cystinosin gene (CTNS). Comparison of longitudinal changes in imaging and dysphagia outcome measures in three interventional groups will help us better characterize changes and potential response to respiratory and swallowing exercises. To achieve this goal, we will assess oral and pharyngeal muscles during swallowing using various imaging methods, gathering baseline data and providing patients with devices to strengthen respective muscle groups before collecting the same data points for comparison one year later. We will also be evaluating possible intervention methods to increase strength in swallowing and respiratory muscles in the same population. This study is led by Dr. Reza Seyedsadjadi.

**The information below tells you about the study that might interest you. You can learn more or speak with the research team to decide if it is a good match for you.**

**Study Name:** Further Characterization of Nephropathic Cystinosis

**What we are studying:** The characterization of muscular involvement in swallowing phases and assessing possible intervention methods to strengthen respiratory and swallowing muscles to improve quality of life.

**Who might qualify:** To participate, you must meet all of the following criteria:

1. You are between the ages of 16 years old and 70 years old

2. You have been previously diagnosed with nephropathic cystinosis and distal myopathy

3. You report or demonstrate dysphagia (difficulty swallowing)

4. You must be able to speak and write in English

**What you will be asked to do in this study:** There will be one secure video conference to assess subject eligibility and to obtain consent. There will be one baseline visit at MGH that will be approximately six hours long, which includes a video fluoroscopy swallow study, ultrasound imaging, walking tests and vital capacity tests. At the end of this visit you will be randomized into one of three treatment groups with a corresponding device. After receiving education on how to use the device, you will be sent home to complete an exercise program in the time in-between visits. You will be required to check in with the study coordinator virtually every three months in-between visits to MGH to ensure no issues have arisen with the program and you are following recommendations. One year after your baseline visit, you will return to MGH to complete an end-of-study visit to repeat all procedures from your baseline visit.

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**To learn more about this study, please contact study coordinator Lauren Politica at lpolitica@mgh.harvard.edu or 617-726-3642.**

Sincerely,

Dr. Reza Seyedsadjadi

*Director, Charcot-Marie-Tooth (CMT) Center of Excellence*

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