



MASSACHUSETTS  
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NEUROLOGY



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## **Distal Myopathy & Muscle Wasting in Nephropathic Cystinosis Research Study**

**About the Study:** Dr. Florian Eichler, the director of the Massachusetts General Hospital (MGH) Center for Rare Neurological Diseases (CRND), is conducting a study to learn more about Distal Myopathy and Muscle Wasting in Nephropathic Cystinosis. The disease progression of Distal Myopathy in Nephropathic Cystinosis (DMNC) varies widely. The myopathy may show variability in the patterns of progression, with some patients displaying more prominent respiratory symptoms and others more dysphagia and swallowing difficulties. This study is currently funded by the Cystinosis Research Foundation (CRF).

### **There are two primary goals of this study:**

1. The study will gather vital information on how cystinosis progresses and how progression varies within the cystinosis population. This information is very useful when determining the effectiveness of therapies in clinical trials. Significant natural history data about the progression of myopathy are lacking and will be crucial in assessing potential therapies for myopathy in cystinosis.
2. It will also identify tools and exercises that can make a difference in patient's lives. We will assess cough airflow measures and implement exercise regimens with the goal to strengthen pulmonary and swallowing functions.

**What the Study Involves:** Participation in this study at Massachusetts General Hospital (MGH) would involve three study visits over two years. One visit will be scheduled during the first year. Two visits will be scheduled during the second year. Each of these visits will involve several different appointments with assessments including: a brief neurological examination, more precise muscle strength evaluation (i.e. dynamometers), a cough airflow study, an assessment of speech and swallowing, and instruction and follow-up for a 5-week Expiratory Muscle Strength Training protocol. 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 from outside of the state of Massachusetts, your travel and hotel expenses will be reimbursed up to \$500. If you are coming from within Massachusetts, we will discuss your individual needs and provide reimbursements if you require hotel accommodation up to \$300. Other travel discounts and funding may be available.

**Benefits of Participating:** Participation in this study will increase our knowledge of the patterns of progression of Distal Myopathy in Nephropathic Cystinosis. Results of all assessments will be made available to patient's home treatment team. As part of the Expiratory Muscle Strength Training Protocol, each participant will receive a high range pressure trainer to use at home.

**Who May Participate:** You are eligible to participate if you are an adult over 18 years of age with a diagnosis of nephropathic cystinosis whether you do or do not have signs of distal myopathy.

**If you would like to participate** in the Distal Myopathy and Muscle Wasting in Nephropathic Cystinosis Study, or if you would like more information, please contact research coordinator Rachel Duong at 617-724-1330 or [rtduong@mgh.harvard.edu](mailto:rtduong@mgh.harvard.edu).