Myopathy and Dysphagia in Adults with Nephropathic Cystinosis

Stacey Sullivan MS, CCC-SLP, Natalie Grant, BA, Feo Price, PhD, Florian Eichler, MD, Reza Sadjadi, MD

Introduction
Nephropathic cystinosis is a lysosomal storage disorder with late-onset systemic complications, such as myopathy and dysphagia. Previously, we evaluated 20 patients over the course of a year to identify sensitive and responsive disease outcomes. Despite mild differences in disability measurements, basic videofluoroscopy identified dysphagia in only 3 patients. Utilizing modern techniques such as MBSImP we showed presence of difficulty in multiple domains including oral and pharyngeal phases of swallowing.

Methods
To further explore potential responsive treatment targets, we repeated MBSImP in 8 of 20 patients with more pronounced swallowing involvement. In addition, we repeated clinical assessments and performed neurophysiology and muscle biopsy to study regenerative capacity of muscle tissue.

Results
We showed significant differences in disability measurements, i.e. Timed Up and Go, Timed 25-Foot Walk, 9- Hole-PEG Test and Grip dynamometry between baseline and follow-up visits ($p<0.05$). Conversely, we observed some reduction in disability related to swallowing in 7 patients. Patients with more advanced dysphagia demonstrated deficit of the pharynx ranging in severity from mildly impaired to limited function of the pharyngeal musculature. We were able to successfully isolate satellite cells from all three muscle biopsies.

Discussion
Repeated exposure to one's own swallowing physiology and resultant function/ deficit over the course of several years via videofluoroscopy and subsequent clinician education sessions following exam may impact ability to volitionally improve function. In this small cohort, this is most evident as related to bolus preparation, transport, and clearance. These are components of swallowing that can be improved with deliberate and focused efforts on behalf of the patient. We could also demonstrate regenerative capacity of the muscle tissue by isolating and culturing satellite cells.