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Clinical myopathy in patients with nephropathic cystinosis

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Abstract

Background
Nephropathic cystinosis is a lysosomal storage disorder. Patient survival years after renal transplantation has revealed systemic complications including distal myopathy and dysphagia.

Methods
We evaluated 20 adult patients with nephropathic cystinosis using patient-reported and clinical outcome measures. Standard motor measures, video fluoroscopy swallow studies, and tests of respiratory function were performed. We also used Rasch analysis of an initial survey to design a 16-item survey focused on upper and lower extremity function, which was completed by 31 additional patients.

Results
Distal myopathy and dysphagia were common in patients with nephropathic cystinosis. Muscle weakness ranges from mild involvement of intrinsic hand muscles to prominent distal greater than proximal weakness and contractures.

Conclusions
In addition to further characterization of underlying dysphagia and muscle weakness, we propose a new psychometrically devised, disease specific, functional outcome measures for distal myopathy in patients with nephropathic cystinosis.