

Medicine (Baltimore). 2005 May;84(3):137-46.

Swallowing dysfunction in 101 patients with nephropathic cystinosis: benefit of long-term cysteamine therapy.

Sonies BC, Almajid P, Kleta R, Bernardini I, Gahl WA.

Oral Motor Function Section, Physical Disabilities Branch, Rehabilitation Medicine Department, Warren Grant Magnuson Clinical Center, National Institutes of Health, Bethesda, MD 20892, USA.

Abstract

Nephropathic cystinosis is a rare, autosomal recessive lysosomal storage disorder caused by mutations in the CTNS gene that codes for a cystine transporter in the lysosomal membrane. Affected patients store 50-100 times the normal amounts of cystine in their cells, and suffer renal tubular and glomerular disease, growth retardation, photophobia, and other systemic complications, including a myopathy and swallowing dysfunction. Using videofluoroscopy and ultrasound examinations, we assessed the swallowing function of 101 patients with nephropathic cystinosis on their most recent admission to the National Institutes of Health Clinical Center between 1987 and 2004. These patients ranged in age from 6 to 45 years; more than half had significant complaints of swallowing difficulty. On examination of barium swallow, the oral, pharyngeal, and esophageal phases of swallowing were abnormal in 24%, 51%, and 73% of patients, respectively. The frequency of dysfunction increased with age for each phase of swallowing. Both the Swallowing Severity Score (a measure of dysfunction on barium swallow) and the Oral Muscle Composite Score (a reflection of vocal strength, oral-facial movement, and tongue and lip function) increased (that is, worsened) with the number of years that a patient was not receiving treatment with cysteamine, the cystine-depleting agent of choice in cystinosis. The severity scores decreased with the number of years on cysteamine therapy. The Swallowing Severity Score varied directly with the severity of muscle disease, but was not correlated with the presence or absence of the 57-kb CTNS deletion that commonly occurs in nephropathic cystinosis patients. We conclude that swallowing dysfunction in cystinosis presents a risk of fatal aspiration, correlates with the presence of muscle atrophy, and, based on cross-sectional data, increases in frequency with age and number of years without cysteamine treatment. Cystine-depleting therapy with cysteamine should be considered the treatment of choice for both pre- and posttransplant cystinosis patients.