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Pulmonary dysfunction in adults with nephropathic cystinosis.

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OBJECTIVE: To characterize the pulmonary dysfunction in patients with nephropathic cystinosis after renal transplantation. DESIGN: Cross-sectional analysis of consecutive adult patients. PATIENTS: Twelve adult, nephropathic cystinosis patients and 3 adult, ocular, nonnephropathic cystinosis patients admitted to the National Institutes of Health Clinical Center. RESULTS: The 12 nephropathic cystinosis patients (age range, 21 to 40 years) showed an extraparenchymal pattern of restrictive lung disease, with inspiratory and expiratory dysfunction. Specifically, the mean FVC was 58% of predicted, the mean FEV(1) was 57% of predicted, and the mean total lung capacity was 66% of predicted, while the mean residual volume was normal. Furthermore, the mean maximal inspiratory pressure for the eight patients tested was 40% of predicted, and the mean maximal expiratory pressure was 26% of predicted. Two patients died of respiratory insufficiency. All the patients had lived at least 17 years, while lacking compliant cystine-depleting therapy with oral cysteamine. Seven patients had a conical chest, restricting excursion, and 10 of the 12 patients had evidence of the myopathy that typifies late cystinosis. In fact, the severity of pulmonary disease correlated directly with the severity of myopathy in our group of 12 patients. In contrast, the lung parenchyma was essentially normal, as gauged by chest radiographs and CT scans of the lung. The three patients with nonnephropathic cystinosis displayed entirely normal pulmonary function. CONCLUSION: The distal myopathy characteristic of nephropathic cystinosis results in an extraparenchymal pattern of restrictive lung disease in adults who have not received long-term cystine depletion. Whether or not oral cysteamine therapy can prevent this complication remains to be determined.

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