Cystinuria and Renal Transplantation

P.	Phil	Tuso
M.	Michelle	Barnett
C.	Chikao	Yasunaga
D.	Donald	Nortman

Transplantation Biology Research Laboratory, Department of Surgery, Cedars Sinai, Medical Center, Beverly Hills, Calif., USA

Phil Tuso, Transplantation Biology Research Laboratory, Department of Surgery, Cedars Sinai Medical Center, 150 N. Robertson Blvd., Suite 250, Beverly Hills, CA 90211 (USA)

Dear Sir,

Cystinuria is a hereditary metabolic disorder involving the transport of the four dibasic amino acids – cystine, lysine, arginine and ornithine. This defect affects primarily the epithelial cells of the renal tubule and the gastrointestinal tract. The defective transport is transmitted as an autosomal recessive trait. Because cystine has a low solubility factor (30(M00 mg/l), these patients form renal calculi when urine output decreases to less than 1 liter/day. Lysine, arginine and ornithine are freely soluble and do not become incorporated in the stones. The plasma levels of these amino acids are either normal or low [1].

Accumulation of the amino acid cystine results in the formation of stones which can cause urinary tract obstruction, infection and ultimately result in end stage renal disease (ESRD). It has been postulated that there is a single enzyme system in the normal renal tubule which is responsible for reabsorption of the four dibasic amino acids [2]. Thus cystinuria is likely not to be a systemic disease, making renal transplantation a good therapeutic option.

There have been at least 4 reported cases of cystine-lysinuria patients who were treated with renal transplantation [3].

Our patient was diagnosed with cystinuria in his early 20s and subsequently developed bilateral staghorn calculi. Despite bilateral pyelolithotomy and creation of bilateral ileal ureters to relieve obstruction, he eventually developed ESRD requiring peritoneal dialysis. The patient eventually underwent a living related transplant without complication. Both urinary cystine excretions and plasma fractionation were monitored prior to and after surgery. Three and one-half years post-renal transplant, urinary amino acid levels remained within normal limits: cystine 37 $\mu\text{mol}/24\text{ h}$, lysine 252 $\mu\text{mol}/24\text{ h}$, ornithine in trace amounts and arginine absent. Renal ultrasound demonstrated no evidence of recurrent stone disease.

The patient presented in this case report and others are examples that a grafted kidney will not be destroyed by recurrent stone disease. Living related donors should be screened for cystinuria and workup should include ultrasound and pyelogram to rule out existing stone disease. We hope that this case will be helpful to others trying to decide whether or not to transplant patients with a history of renal loss secondary to cystinuria.

References

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