Successful living-related kidney transplantation in hereditary renal hypouricaemia

Sir,

Hereditary renal hypouricaemia (MIM: 220150) is a syndrome that involves a defect in urate transporter1 (URAT1) for urate reabsorption at the brush border membrane of the proximal tubule in the kidney [1]. A G774A mutation in SLC22A12 encoding URAT1 is the dominant mutation in Japanese [2]. Since hypouricaemia itself does not induce any symptoms, hereditary renal hypouricaemia is sometimes overlooked. Exercise-induced acute renal failure and urolithiasis are two major complications affecting renal function [3,4]. The short-term prognosis of exercise-induced acute renal failure is good, while the long-term prognosis is not known [5].

We report a kidney transplantation between relatives with hereditary renal hypouricaemia. A 29-year-old man, who received a living-related renal graft at the age of 24 years (in August 1994) from his father, was referred to our hospital in July 1999 for maintenance post kidney transplantation. We confirmed a well-functioning kidney graft (serum creatinine 1.3 mg/dl, creatinine clearance 69 ml/min) with no proteinuria or haematuria. Nevertheless, he had hypouricaemia at 1.0 mg/dl with a high fractional excretion of uric acid (uric acid clearance/creatinine clearance) at 57.9% (normal range, 6–10%) and a normal urinary urate excretion of 0.40 mg/kg/h. We diagnosed him as having hereditary renal hypouricaemia. Both the recipient and donor had the same disorder of urate metabolism and were homozygous for G774A. However, they had not been informed about renal hypouricaemia at the time of kidney transplantation. The patient did not show hypouricaemia during maintenance haemodialysis because an anuric dialyzed patient with hereditary renal hypouricaemia does not eliminate uric acid in the urine. In addition, the renal hypouricaemia had been overlooked in the donor. Fortunately, the patient has had an excellent outcome, 11 years after his kidney transplantation with no major complications, such as renal stones or exercise-induced acute renal failure.

In our case, both donor and recipient were at high risk for complications. This case demonstrates that we need not shrink from kidney transplantation in hereditary renal hypouricaemia. However, we should take care that renal transplantation patients are aware of all the risks involved. If patients are found to have hereditary renal hypouricaemia, we should direct them to avoid hard exercise, drink plenty of water, and alkalinize their urine [3,4]. We need to remember that an anuric dialyzed patient with hereditary renal hypouricaemia does not have hypouricaemia.

Conflict of interest statement. None declared.

Division of Kidney and Hypertension, Izumi Yamamoto
Department of Internal Medicine, Hiroyasu Yamamoto
The Jikei University School of Medicine, Kimiyoshi Ichida
Tokyo, Jun Mitome
Japan, Naohiko Katoh
Email: izumi26@jikei.ac.jp, Keitaro Yokoyama
Tatsuo Hosoya


doi:10.1093/ndt/gfk103