Letter to the editor

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Successful renal transplantation in a patient with familial lecithin: cholesterol acyltransferase deficiency

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Sir: Familial lecithin: cholesterol acyltransferase (LCAT) deficiency, first reported by Gjone and Norum [3], is clinically characterized by diffuse corneal opacities, normochrome anemia, and proteinuria, progressing to renal insufficiency. Lipid deposits are found in the cornea, erythrocytes, spleen and kidneys. All plasma lipoproteins are abnormal and all individual fractions have a higher amount of free cholesterol than is considered normal. Triglycerides are usually elevated and lysolecithin low.

Treatment with low-fat food or hypolipemics does not seem to prevent the development of renal failure [4]. Renal transplantation is therefore indicated when kidney failure occurs in patients with familial LCAT deficiency [1, 2].

We report on a 28-year-old patient from Bulgaria, afflicted with familial LCAT deficiency. In 1975 he had proteinuria. In 1977 he was admitted to the hospital with nephrotic syndrome, hepatosplenomegalia, hypertension, and annular corneal opacities. Renal failure started in 1981. Renal biopsy showed deposits of amorphous material in the glomerular capillaries and some foam cells. His 4 year younger sister is also afflicted to a lesser degree. Our patient started hemodialysis in 1985. Due to arterial hypertension and ocular lipid deposits, he had several complications, namely hemorrhage and retinal detachment, and he lost his eyesight.

In February 1987 the patient was given a cadaveric kidney transplant with four HLA A, B and DR incompatibilities. Before transplantation his ASAT and ALAT were elevated. He received triple immunosuppressive treatment with cyclosporin to a mean whole blood level of 410–190 ng/ml (Sandoz RIA kit), prednisolone, and azathioprine. During the first month we noticed an improvement in his transaminases. Up until June 1987 he remained well but his renal function was imperfect, with a serum creatinine level of 190–257 μmol/l. In July the patient was hospitalized at Hôpital Necker (Paris) with a serum creatinine level of 204–188 μmol/l, blood urea 16–17 mmol/l, hemoglobin 11.7 g/l, hyperuricemia 600 mmol/l, cholesterol 2.6–3.4 mmol/l, triglycerides 6.65 mmol/l, and HDL 0.07–0.09. The transplant biopsy showed tubulointerstitial rejection changes and discrete segmental changes compatible with his metabolic disease. After high-dose steroid treatment his creatinine level decreased to 140–150 μmol/l.

Four years after renal transplantation our patient is well and is receiving triple immunosuppressive therapy. He is normotensive, his creatinine level is 130 μmol/l, and he has a minimal proteinuria of 200 mg/24 h. The transplant biopsy shows some foam cells and lipid deposits in the glomerular capillaries.

In this report of renal transplantation in a patient with familial LCAT deficiency, coming from a family with two afflicted members, it is interesting to note that the transaminases returned to normal values after transplantation and immunosuppressive treatment.

One may conclude that familial LCAT deficiency does not contraindicate renal transplantation. The metabolic abnormalities frequently affect the transplant, yet it is possible that triple immunosuppressive therapy (cyclosporin + azathioprine + steroids), in combination with fat restriction, could prevent the rapid recurrence of the specific changes in the transplanted kidney.

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