Sir,

Lecithin: cholesterol acyltransferase (LCAT) deficiency is a rare familial disease characterized by absence of esterification of cholesterol in plasma. This results in abnormality of virtually all plasma lipoproteins [1]. Deposition of the abnormal lipids in kidney, cornea, erythrocytes and vessel walls is believed to be the basis of the clinical sequelae of the disease.

We measured plasma beta-2-microglobulin (Phadebas RIA kit) and urinary albumin by radioimmunoassay [2] in 2 patients with LCAT deficiency [3]. One of them (S. F.) has a definite, biopsy-proven renal lesion and proteinuria (5–10 g/day) while the other (D. H.) has normal renal function. Both patients have had repeatedly normal plasma creatinine and creatinine, inulin and urea clearances over 5 years of observation. However, beta-2-microglobulin was 3 and 3.8 mg/l in S.F. (normal 1.1–2.4 mg/l) on two occasions over the last 6 months. A normal level of this protein was found in D.H. Urinary albumin: creatinine ratio was between 2.7 and 4.8 in S.F. on five separate examinations and 0.06–0.067 on three occasions in D.H. (normal 0.003–0.013).

Two conclusions can be drawn from these observations. First, in agreement with others [4] we can state that beta-2-microglobulin appears to be a better indicator of renal damage than the other functional tests (in the patient S. F.). Secondly, the albumin: creatinine ratio (and the absolute excretion of albumin) is increased in the asymptomatic patient with LCAT deficiency (D. H.), confirming the possibility that she, similarly to a recently discovered Irish patient with LCAT deficiency without functional renal involvement [Dr. G. Thompson, personal commun.], may have glomerular lipid deposits affecting the albumin losses. Measurement of urinary albumin by radioimmunoassay is a useful and sensitive indicator of renal function [5, 6]. Both of these tests suggest abnormality of renal function in our patients.

References
