A 42-year-old Japanese man visited our nephrology department after proteinuria without hematuria was detected in a medical checkup. A physical examination showed no abnormalities. His renal function was normal (serum creatinine concentration: 0.65 mg/dL). The urinary analysis at the first visit revealed fat bodies with a whorl-like appearance; so-called mulberry bodies (Picture 1). These bodies showed a characteristic Maltese Cross configuration under polarized light microscopy (Picture 2), which was highly suggestive of Fabry disease (1). Indeed, the alpha-galactosidase (GLA) activity in the patient’s white blood cells was as low as 0.1 nmol/h/mg and his serum globotriaosylceramide level was elevated to 69 nmol/L. A genetic analysis revealed a thymine insertion in exon 5 of the GLA-gene, which confirmed the diagnosis of the classic Fabry disease phenotype (2). In a further examination, left ventricular hypertrophy and whorl-like corneal opacity were observed.

In the present case, the detection of mulberry bodies in a microscopic urinalysis was the clue to the diagnosis. Enzyme replacement therapy was initiated four months after the patient’s diagnosis.

The authors state that they have no Conflict of Interest (COI).

References


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