Fabry Disease Diagnosed Based on the Detection of Urinary Mulberry Bodies

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Picture 1.

Picture 2.

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).

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