中文題目：罕見法布瑞式症以不明熱為最初表現:病例報告

英文題目： Fabry disease: a rare cause of fever of unknown origin

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Introduction:

Fever of unknown origin (FUO) is a clinical scenario of diverse etiology, yet 10-15% cases are idiopathic. Various diagnostic modalities exist in our armamentarium against FUO, but renal biopsy is rarely enlisted in the literature. Here we describe a young man with Fabry disease and an FUO which subsided after enzyme replacement therapy.

Case presentation and management:

A 32-year-old Asian male presented with a year-long intermittent fever. Fever was diurnal and relieved with acetaminophen. He denied any systemic disease or limb pain. Physical examination was unremarkable and without angiokeratomas. Serum creatinine was 0.9 mg/dl (79.5μmol/L, eGFR 104 ml/min/1.73 m^2 by MDRD Study formula), abdominal computed tomography was negative and urinalysis revealed proteinuria (1.5-2.5g/day) without hematuria. He was treated with prednisone 10 mg and cyclosporine 25 mg daily for six months, but fever and proteinuria persisted, and he developed avascular necrosis of the femoral head. A renal biopsy disclosed abundant "zebra bodies" in the podocyte. Fabry disease was confirmed by gene analysis and reduced blood leukocytes α-galactosidase A activity. Several months after enzyme replacement therapy his fever resolved, but proteinuria remained unchanged.

Discussion:

Fabry disease is first described in 1898 by dermatologists Johannes Fabry and William Anderson, but the exact cause has not been delineated until 1970s, when α -galactosidase A deficiency is pinpointed as origin. It is frequently misdiagnosed as rheumatologic syndrome, but is not listed as one of the major culprits of FUO. Hyperthermia due to anhydrosis from sympathetic fiber neuropathy and sweat gland deposition would not be expected to be diurnal nor suppressed by acetaminophen, while fever due to inflammation would be expected to be diurnal and suppressed by acetaminophen. The explanation for fever in this case is not apparent, but inflammation has been reported in association with Fabry disease.

Conclusion:

This case demonstrates the potential role of kidney biopsy in the diagnosis of Fabry

disease presenting with proteinuria and FUO. In light of the presentation in our case, delay in diagnosis and empiric treatment with immunosuppressive agents can be associated with potentially avoidable complications.