KSN 2018
Seoul, Korea · May 17 - 20

Abstract Type : Poster
Presentation No. : POT 015

Monitoring globotriaosylsphingosine before and after enzyme replacement therapy in a patient with Fabry disease: A case report

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Case Study: Fabry disease is an X-linked lysosomal storage disorder caused by deficiency of α-galactosidase A activity. It leads to dysfunctions in multiple organs, including the kidney, heart, and brain. Representative deposits are globotriaosylceramide (Gb3) and globotriaosylsphingosine (lyso-Gb3). Recent studies have proposed lyso-Gb3 to be a therapeutic biomarker. Herein, we report a case in which lyso-Gb3 was traced before and after enzyme replacement therapy in a patient with Fabry disease. A 29-year-old man was diagnosed with Fabry disease on the basis of reduced α-galactosidase A enzyme activity in leukocytes (3.3 nmol/h/mg protein) and α-galactosidase A gene mutation (Asp92Gly). At baseline, the patient had angiokeratoma, decreased kidney function (serum creatinine, 1.90 mg/dl; estimated glomerular filtration rate, 46.6 ml/min/1.73 m^2), proteinuria (random urine protein-to-creatinine ratio, 3.1 g/g), cardiac basal septal wall hypertrophy (14 mm), and a suspected ischemic lesion in the right posterolateral putamen (6-mm-high signal intensity on a T2-weighted-Fluid-Attenuated Inversion Recovery image, and low intensity on a T1-weighted brain magnetic resonance image). A biopsied specimen from the kidney showed diffuse lipid deposits in podocytes, glomerular endothelial cells, and mesangial cells. The patient was treated biweekly with agalsidase beta (Fabrazyme®), and both plasma Gb3 and lyso-Gb3 levels were estimated every 3 months. Before enzyme replacement therapy, the plasma Gb3 and lyso-Gb3 were 10.9 μg/ml (reference range, 3.9–10.9 μg/ml) and 108.0 ng/ml (reference range, ≤1.74 ng/ml), respectively. After 6 months of treatment, the levels of Gb3 and lyso-Gb3 dropped to 6.4 μg/ml and 18.7 ng/ml, respectively. During the treatment period, kidney function did not decrease further (serum creatinine, 1.81 mg/dl; random urine protein-to-creatinine ratio, 2.2 g/g). The present case suggests that lyso-Gb3 can be useful in monitoring the response to enzyme replacement therapy.