S21-4 Measurement of plasma lyso-Gb3 as a new marker for Fabry disease

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Fabry disease is an X-linked genetic disorder caused by a deficiency of α-galactosidase A (GLA) activity. As enzyme replacement therapy (ERT) involving recombinant GLAs has been introduced for this disease, a useful biomarker for diagnosis and monitoring of therapy is strongly required. We measured globotriaosylsphingosine (lyso-Gb3) in plasma samples from hemizygous males and heterozygous females with Fabry disease, and investigated the response of plasma lyso-Gb3 in a male Fabry patient who had undergone ERT for 4 years to determine whether plasma lyso-Gb3 could be a biomarker of Fabry disease. The results revealed that plasma lyso-Gb3 was apparently increased in male patients. In female patients, plasma lyso-Gb3 was moderately increased in both symptomatic and asymptomatic cases, and there was a correlation between the increase in lyso-Gb3 and the decrease in GLA activity. The plasma lyso-Gb3 in the Fabry patient who had received ERT was elevated at baseline and fell impressively on ERT. Plasma lyso-Gb3 could thus be a potential biomarker of Fabry disease. We are trying to determine the consentration of lyso-Gb3 in Fabry mice tissues.