Urinary titin as a biomarker in Fukuyama congenital muscular dystrophy

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Abstract
Fukuyama congenital muscular dystrophy (FCMD) is the second most prevalent childhood-onset muscular dystrophy in Japan. It is an autosomal recessive disorder caused by the fukutin mutation (FKTN), characterized by muscle wasting and brain abnormalities. So far, serum creatine kinase (CK) is recognized as the only biomarker for FCMD. Recently, an ELISA assay to quantify the N-terminal fragment of titin in urine was developed. Urinary titin concentration is elevated in patients with Duchenne muscular dystrophy (DMD) compared to normal controls. Levels vary according to age with excellent sensitivity and specificity for detecting DMD, and they can be used as a diagnostic and disease progression marker. In this study, we measured the urinary titin concentration of 18 patients with FCMD. It was remarkably higher than normal controls and correlated with CK. Especially in homozygotes, the score for gross motor function measure, which is a quantitative motor scale for FCMD, was correlated with urinary titin concentration. Elevated urinary titin concentrations were thought to be reflective of a common pathophysiology with DMD. Urinary titin concentrations can assist with making the diagnosis of FCMD and to estimate the patient's motor function at that point.

Keywords: Fukuyama congenital muscular dystrophy; Gross motor function measure; Urinary titin concentration.

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