The gross motor function measure is valid for Fukuyama congenital muscular dystrophy.

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**Abstract**

Fukuyama congenital muscular dystrophy (FCMD) is the second most common muscular dystrophy in Japan. FCMD is an autosomal recessive disorder caused by mutations in the fukutin gene. The main features of FCMD are a combination of infantile-onset hypotonia, generalized muscle weakness, eye abnormalities, and mental retardation associated with cortical migration defects, and most patients are never able to walk. To date, the development of a quantitative motor scale for FMCD has been difficult due to the moderate-to-severe intellectual impairment that accompanies FCMD. Gross motor function measure (GMFM), originally developed as a quantitative motor scale for cerebral palsy, can precisely and quantitatively assess motor function without complicated instructions, and was recently reported to be useful in the assessment of Down syndrome and spinal muscular atrophy. To confirm the validity of GMFM for the assessment of FCMD, 41 FCMD patients (age range: 0.6-24.4 years) were recruited for this study. The GMFM scores correlated significantly with those of two previously used motor scales, and the time-dependent change in GMFM scores was consistent with the natural course of FCMD. The inter-rater reliability, based on determinations made by four physiotherapists blinded to each other’s assessment results, was excellent. We concluded GMFM to be a useful and valid measure of motor function in FCMD patients.

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**KEYWORDS:** Fukuyama congenital muscular dystrophy; Gross motor function measure; Intellectual involvement

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