National registry of patients with Fukuyama congenital muscular dystrophy in Japan.

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Abstract

Fukuyama congenital muscular dystrophy (FCMD) is the second most common form of muscular dystrophy in the Japanese population and is caused by mutations in the fukutin (FKTN) gene. In 2011, the Japan Muscular Dystrophy Association (JMDA) developed a nationwide registry of genetically confirmed patients with FCMD. We retrospectively reviewed the registry dataset of patients with FCMD to obtain data, including age, sex, developmental milestones, intellectual level, complications, and primary treatments. In total, 207 patients with FCMD (104 boys and 103 girls) were registered by the end of September 2013. Mean patient age at first registration was 8.1±7.8 years (median, 6 years; range, 0-42 years). A homozygous 3-kb founder insertion mutation in the FKTN gene was present in 80% of registrants, whereas 20% had a compound heterozygous mutation. Sixty-nine patients (33%) had febrile seizures and/or epilepsy. Myopia was the most frequently detected abnormality (8.7%), followed by strabismus (5.9%). Overall, 16% of patients required respiratory support and this percentage increased with age. Cardiac dysfunction was detected in 16%, and dysphagia was observed in 22% of patients with FCMD. The FCMD patient registry is useful for clarifying the natural history of FCMD and recruiting patients for clinical trials.

KEYWORDS: FKTN; Fukuyama congenital muscular dystrophy; Japan Muscular Dystrophy Association (JMDA); Natural history; Patient registry

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