Abstract
Fukuyama congenital muscular dystrophy (FCMD), the second most common muscular dystrophy in the Japanese population, is an autosomal recessive disorder caused by mutations in the fukutin (FKTN) gene. The main features of FCMD are a combination of infantile-onset hypotonia, generalized muscle weakness, eye abnormalities and central nervous system involvement with mental retardation and seizures associated with cortical migration defects. The FKTN gene product is thought to be necessary for maintaining migrating neurons in an immature state during migration, and for supporting migration via α-dystroglycan in the central nervous system. Typical magnetic resonance imaging findings in FCMD patients are cobblestone lissencephaly and cerebellar cystic lesions. White matter abnormalities with hyperintensity on T(2)-weighted images are seen especially in younger patients and those with severe phenotypes. Most FCMD patients are mentally retarded and the level is moderate to severe, with IQs ranging from 30 to 50. In our recent study, 62% of patients developed seizures. Among them, 71% had only febrile seizures, 6% had afebrile seizures from the onset, and 22% developed afebrile seizures following febrile seizures. Most patients had seizures that were controllable with just 1 type of antiepileptic drug, but 18% had intractable seizures that must be treated with 3 medications.

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Seizure-genotype relationship in Fukuyama-type congenital muscular dystrophy.

Characteristics of neurons and glia in the brain of Fukuyama type congenital muscular dystrophy.
Review article

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