[Clinical features and FKRP mutations of congenital muscular dystrophy 1C].

[Article in Chinese]
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Abstract
Congenital muscular dystrophy type 1C (MDC1C) is caused by the homozygous or compound heterozygous mutations of the FKRP gene. This article reported the clinical and mutation features of a child with MDC1C. The boy aged 8 months visited the hospital due to delayed development. As for clinical manifestations, the boy could not turn over or sit stably by himself, and there was a significant reduction in muscle tension; biceps reflex in both upper extremities and patellar tendon reflex and Achilles tendon reflex in both lower extremities could not be induced. The boy also had a stereotyped facial expression and strabismus. Gene detection revealed c.350C>G and c.1303C>T compound heterozygous mutations in the FKRP gene. The c.350C>G mutation came from the mother and had been reported as a pathogenic missense mutation. The c.1303C>T mutation came from the father and was a new missense mutation, and a bioinformatics analysis showed that it might be a pathogenic mutation. The boy was diagnosed with MDC1C with reference to the clinical features of hypotonia and motor developmental delay and FKRP gene mutation sequencing.

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