[Analysis of POMT1 gene mutation in a pedigree affected with congenital muscular dystrophy].


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Citation

Abstract
OBJECTIVE To analyze mutation of POMT1 gene in a Chinese family affected with congenital muscular dystrophy (CMD). METHODS Peripheral blood samples of the family including one affected and two unaffected individuals, in addition with chorionic villous sample from the fetus, were collected. PCR was used to amplify exons 19 and 20 of the POMT1 gene, and the products were sequenced directly. Based on the result of genetic testing, prenatal diagnosis of the fetus was attained. RESULTS The proband was found to carry a heterozygous missense mutation c.1939G>A (p.Ala647Thr) in exon 19 of the POMT1 gene inherited from the mother and a heterozygous frameshift mutation c.2141delG (p.Trp714Ter) in exon 20 inherited from the father. Prenatal diagnosis revealed that the fetus has carried
the c.1939G>A (p.Ala647Thr) missense mutation. With the disease causing mutation, the fetus was predicted to have similar phenotype as its mother. CONCLUSION The compound heterozygous mutations of c.1939G>A (p.Ala647Thr) and c.2141delG (p.Trp714Ter) probably underlie the CMD in this family. Based on the result, prenatal diagnosis may be provided.

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