Idiopathic hyperCKemia and malignant hyperthermia susceptibility.

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Abstract

PURPOSE: HyperCKemia is a persistent rise in serum creatine kinase (CK) levels of at least 1.5 times the normal value, as evidenced by a minimum of two measurements at 30-day intervals. One of the neuromuscular diseases associated with hyperCKemia is malignant hyperthermia (MH). This study investigated the susceptibility to MH in patients with hyperCKemia via in vitro contracture testing (IVCT) and a search of mutations in the RYR1 gene.

METHODS: Patients in an MH centre were followed from 1997-2012, and their epidemiologic, clinical, and laboratory data were analyzed, including IVCT, muscle histochemical analysis, and next-generation sequencing molecular analysis.

RESULTS: There were nine patients (eight male) in our study with a mean (SD) age of 33 (12) yr. Four patients were Caucasian and five were African Brazilian. Most complained about myalgia or cramps, but all had a normal neurological examination. They persistently presented with hyperCKemia from three months to ten years, with a mean (SD) CK value of 788 (507) IU·L\(^{-1}\) ranging from 210-1,667 IU·L\(^{-1}\). These values corresponded to a 1.5- to nine-fold increase in the normal value (mean increase, 3.7-fold). Six patients were MH susceptible (MHS) after a positive IVCT. Histopathological muscular analysis disclosed unspecified changes in four of the MHS patients. Mitochondrial proliferation was observed in the other two MHS patients and in three MH negative patients. No pathogenic mutations were identified in the RYR1 gene in the five patients evaluated.

CONCLUSION: When investigating patients with idiopathic hyperCKemia, susceptibility to MH should be taken into account, and guidance should be offered to prevent anesthetic complications in the family.

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