

Malignant Hyperthermia and *Ryanodine Receptor Type 1* Gene (*RyR1*) Mutation in a Family in Singapore

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Abstract

Introduction: Sporadic clinical episodes of malignant hyperthermia (MH) that develop during general anaesthesia (GA) have been reported in Singapore. However, there is no published local report of a confirmed case of MH susceptibility (MHS) by skeletal muscle contracture tests and/or molecular tests. **Materials and Methods:** We report 2 patients from an extended family who developed signs of clinical MH while under GA. The MH episodes were successfully treated with intravenous dantrolene sodium. Sequence analysis of the entire *Ryanodine Receptor Type 1* (*RyR1*) coding gene was carried out in an index patient. **Results:** The index patient was found to carry a c.7373G>A (p.Arg2458His) mutation in exon 46. This particular mutation satisfies the criteria for a MHS causative mutation. Hence, the index patient was considered to be MHS and did not need to undergo further muscle contracture testing. The same mutation was also found in 3 other members of his extended family. **Conclusion:** This is the first report of a Singaporean family with at least 4 members carrying a MH-causative mutation in *RyR1* gene. This report serves to highlight the existence of the putative gene for MH in Singapore, and the need for clinical vigilance during anaesthesia involving the use of triggering agents.

Ann Acad Med Singapore 2017;46:455-60

Key words: Dantrolene, Inhalational agent, Suxamethonium

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