Case Study – Malignant Hyperthermia

Names and other identifying information have been changed to protect patient confidentiality.

Chad, 39 year old male

Chad has never had any serious health conditions or surgeries. His ActX testing showed that he had a variant in the gene RYR1 which is associated with Malignant Hyperthermia susceptibility (MHS). Patients with MHS who undergo general anesthesia using inhaled, volatile anesthetics such as sevoflurane, isoflurane, desflurane, or enflurane are at risk of developing malignant hyperthermia, a life threatening condition, usually on the operating room table.

In malignant hyperthermia, a failure in calcium balance regulation leads to the production of excess body heat and lactate, which in turn leads to serious complications and high mortality rate if left untreated. About 1 person out of 2000 carries an RYR1 variant pre-disposing them to malignant hyperthermia. Variants in CACNA1S can also predispose to malignant hyperthermia.

Outcome

Knowing that he carries a variant for malignant hyperthermia, Chad and his physicians can make sure that a different type of anesthetic is used during any future surgery.

Additional information on Malignant Hyperthermia

The RYR1 (Skeletal Muscle-Type Ryanodine Receptor 1) protein plays an important role in encoding for skeletal muscle receptors, which regulate calcium balance. Administration of inhaled, volatile anesthetics can trigger a release of calcium stores, which can lead to muscle contracture, glycogenolysis, and increased cellular metabolism. The combination of this variant with the administration of inhaled anesthetics may trigger a production of excess body heat and lactate, which can lead to serious complications and high mortality rate if left untreated.

References

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