Malignant Hyperthermia: testing

Testing for malignant hyperthermia largely falls into two “buckets”. The “gold standard” (if it can be called that) is the caffeine-halothane contracture test (CHCT) or in vitro contracture test (IVCT) depending on if you’re on team North America or team Europe. This involves testing live tissue via muscle biopsy which requires the patient to travel to 1 of 5 (in North America) approved sites adding extra cost to an already costly test. The tests are quite sensitive (97-99%) but less specific (78-94%) resulting in a high incidence of false positives: about 1 in 5. However, given the sensitivity, false negatives in contracture tests are rare.

More commonly, genetic testing is performed looking for *RYR1* mutations on chromosome 19 and mutations associated with the *CACNA1S* and *STAC3* genes. Although only a few labs are able to perform the test, this is far cheaper and requires only a blood sample which can be mailed. Although over 400 RYR1 variants associated with MH have been discovered, sensitivity for genetic testing remains 60% making it impossible to rule out a patient’s susceptibility to malignant hyperthermia.

Further Reading:

Ellinas H, Albrecht MA. Malignant Hyperthermia Update. Anesthesiol Clin. 2020 Mar;38(1):165-181. doi: 10.1016/j.anclin.2019.10.010. PMID: 32008650.

Rosenberg H, Pollock N, Schiemann A, Bulger T, Stowell K. Malignant hyperthermia: a review. Orphanet J Rare Dis. 2015 Aug 4;10:93. doi: 10.1186/s13023-015-0310-1. PMID: 26238698; PMCID: PMC4524368.