Your CACNA1S Genetic Test Results and What They Mean

CACNA1S: Increased Risk for Malignant Hyperthermia

Pharmacogenomic Testing Overview

Pharmacogenomic (PGx) testing looks at how your genes affect your response to certain medications. Genes are pieces of DNA that provide instructions to make our bodies look and work as they do. Some genes affect the way medications work in the body. When comparing a group of people, there can be slight differences in the structure of each person's genes. These differences can affect how people respond to medications.

Some gene differences might make it harder for the body to get rid of some medications. This means that the usual dose of the medication may cause unexpected side effects. Some gene differences can cause the body to use up a medication too fast. This means that normal doses will not work as well, and the person may need higher doses. Some gene differences will not let certain medications work in the body at all. This means a different medication may work better. Some gene differences increase your chances of side effects to medications. This means that you may need to avoid certain medications.

This gene test may have been part of a panel of genes or a single gene test. The results and affected medications described below may not be relevant to your current care, but could be in the future.

About the CACNA1S Gene

The test we did was for a gene called the calcium voltage-gated channel subunit alpha1 S (abbreviated CACNA1S). Variations in the CACNA1S gene can impact how our body responds to certain anesthesia and neuromuscular blocker medications given during surgery. Depending on these variations, people are considered to be at Normal Risk or Increased Risk for Malignant Hyperthermia. Malignant Hyperthermia is a type of severe reaction to anesthesia where your body temperature increases dangerously high, which can be fatal if not treated immediately. CACNA1S is one of two genes we currently know of that impacts your risk of developing malignant hyperthermia, with the other being the RYR1 gene. If indicated for surgical anesthesia, your healthcare provider can analyze the results of these genes and additional factors when determining what anesthesia medications to choose.

Your CACNA1S result puts you in the increased risk for malignant hyperthermia group. In people who have increased risk for malignant hyperthermia, they should avoid the use of the following anesthesia and neuromuscular blocker medications – desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, and succinylcholine. Your healthcare provider can look at other clinical factors before deciding what anesthesia agents to use. Other family members (parents, siblings, future children, etc.) should also undergo testing for malignant hyperthermia susceptibility variants in the CACNA1S and RYR1 genes.

The following medications interact with CACNA1S:

Anesthetics (used to provide pain relief during surgeries or medical procedures): desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane
Neuromuscular blocker (helps relax muscles before procedures): succinylcholine

Do not make any adjustments to your medications without first speaking to your healthcare provider.

Because your genes stay the same even as you age, it is important for you to share this result with your other doctors and pharmacists outside Children's Mercy. This result may affect how doctors prescribe medications throughout your life.

More Information

- Research continues to be done on what medications are affected by genetic test results. For more details about CACNA1S and malignant hyperthermia, please go to https://www.mhaus.org/patients-and-families/learn-about-mh/ or www.clinpgx.org.
- If you have questions about your pharmacogenetic test results or specific treatment options, discuss them with your healthcare provider or call 816-601-3360 to schedule an appointment at the Children's Mercy GOLDILOKs Clinic.
- If interested in volunteering for pharmacogenetic research, please contact the Children's Mercy Research Institute at pharmacogeneticsresearch@cmh.edu.

Revised October 2025