



Your G6PD Genetic Test Results and What They Mean

G6PD: Deficient Function with CNSHA

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. 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 G6PD Gene

The test we did was for a gene called Glucose-6-Phosphate Dehydrogenase (abbreviated G6PD). This gene makes an enzyme that is involved in a number of different cellular metabolism processes. How well this enzyme functions can impact your risk of side effects to certain medications, specifically the side effect of hemolytic anemia. Hemolytic anemia is a condition where more red blood cells are being destroyed than produced. Hemolytic anemia can cause you to feel very tired, dizzy, and affect your heart rate and breathing. This may mean you need lower doses or to avoid certain medications. It is common to have slight variations in the G6PD gene that affect how the enzyme works. Depending on these variations, people are considered to have Deficient, Deficient with CNSHA (chronic non-spherocytic hemolytic anemia), Normal, or Variable Function.



Your G6PD result puts you in the deficient function with CNSHA group. People who have deficient function for G6PD with CNSHA chronically have hemolytic anemia and experience intermittent exacerbations when exposed to certain triggers, such as medications. You need to avoid certain medications that interact with G6PD due to the increased risk of severe acute exacerbations of hemolytic anemia when exposed to these medications. Your healthcare providers can use your test result to help decide what medications and what dose may be the best for you.

The following medications interact with the G6PD enzyme:

Antibiotics (used to treat and prevent certain infections): dapson, nitrofurantoin, primaquine, tafenoquine

Other: methylene blue (used to treat a blood disorder that affects how oxygen is transported), pegloticase (used to treat gout), rasburicase (used to treat high levels of uric acid in the blood), toluidine blue (used to help identify cancer affected tissue)

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 the G6PD gene, please go to 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.

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