



Your CFTR Genetic Test Results and What They Mean

CFTR Kalydeco (ivacaftor) Responsive

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.

About the CFTR Gene

The test we did was for a gene called the Cystic Fibrosis Transmembrane Conductance Regulator (abbreviated CFTR). This gene makes a protein in your cell membranes involved in a number of vital processes to help our bodies function normally. In patients with cystic fibrosis, genetic variation in the CFTR gene leads to this protein not functioning properly. If untreated, this can lead to a number of complications. These complications include thickened mucus in the lungs, respiratory infections, and pancreatic insufficiency. Certain genetic variations make you more or less likely to respond to medications used to treat cystic fibrosis. Your CFTR result helps your healthcare providers predict how you will respond to the following cystic fibrosis medications: Alyftrek (vanzacaftor/tezacaftor/deutivacaftor), Kalydeco (ivacaftor), Orkambi (ivacaftor/lumacaftor), Symdeko (tezacaftor/ivacaftor), and Trikafta (elexacaftor/tezacaftor/ivacaftor).

Your CFTR result puts you in the Kalydeco (ivacaftor) responsive group. Based on your genetic results for CFTR, you are likely to respond well to Kalydeco, which is one of many medications that can be used to treat and manage cystic fibrosis. Your healthcare providers can



use this test result and other clinical factors, like your age and weight, to help decide what medications and what dose may be the best for you.

The following medications interact with the CFTR protein:

Cystic Fibrosis medications: Alyftrek (vanzacaftor/tezacaftor/deutivacaftor), Kalydeco (ivacaftor), Orkambi (ivacaftor/lumacaftor), Symdeko (tezacaftor/ivacaftor), Trikafta (elexacaftor/tezacaftor/ivacaftor)

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 CFTR 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 816-983-6490 to schedule an appointment at the Children's Mercy Cystic Fibrosis Care Center.
- If interested in volunteering for pharmacogenetic research, please contact the Children's Mercy Research Institute at pharmacogeneticsresearch@cmh.edu.

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