



Your TPMT Genetic Test Results and What They Mean

TPMT: Indeterminate Metabolizer

Pharmacogenomic Testing Overview

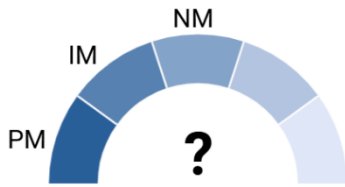
Pharmacogenomic (PGx) testing looks at how your genes affect your response to certain medications. Genes are pieces of DNA that provide instructions to make our bodies look and work as they do. Some genes affect the way medications work in the body. When comparing a group of people, there can be slight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

The test we did was for a gene called Thiopurine Methyltransferase (abbreviated TPMT). This gene makes an enzyme that breaks down, or metabolizes, medications in the body. The breakdown of thiopurines is also impacted by another gene, so your healthcare provider can also analyze your NUDT15 gene result before determining what dose to choose. It is common to have slight variations in the TPMT gene that affect how the enzyme works. Depending on these variations, people are considered Poor (PM), Intermediate (IM), Normal (NM), or Indeterminate Metabolizers.



Your TPMT result puts you in the indeterminate metabolizer group. In people who are indeterminate metabolizers, the TPMT enzyme function is not known. Certain genetic variants are rare, so we do not currently know how the TPMT enzyme functions in all patients. About 2% of our patients are TPMT indeterminate metabolizers. In the future as more research is done, we may be able to assign you to one of the TPMT metabolizer groups shown in the figure. Your healthcare

provider can also look at your NUDT15 gene test result before deciding what dose of a thiopurine medication you might need.

The following are medications metabolized by the TPMT enzyme:

Thiopurines (used to treat cancer and autoimmune diseases): azathioprine, mercaptopurine, thioguanine

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 which medications are broken down by TPMT, 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 GOLDILOCKS Clinic.
- If interested in volunteering for pharmacogenetic research, please contact the Children's Mercy Research Institute at pharmacogeneticsresearch@cmh.edu.

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