



# Your *TPMT* Genetic Test Results and What They Mean

*TPMT* : Uncertain #305



# Common differences in the *TPMT* gene can affect how you respond to some medicines

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We recently tested you for the *TPMT* gene. This info sheet explains the test, your results, and what your doctor may do with that information.

Genes are pieces of DNA that provide instructions to make our bodies look and work as they do. Some genes affect the way medicines work in the body. When comparing a group of people, there can be slight differences in each gene's structure. These differences can affect how people respond to medicine.

Some gene differences might make it harder for the body to get rid of some medicines. This means that usual doses of the medicine could give some people unexpected side effects. Some gene differences can cause the body to use up a medicine too fast. This means that normal doses won't work as well and the person may need higher doses. Some gene differences won't let certain medicines work in the body at all. This means a different medicine may work better.

**What we tested:** We tested a gene called Thiopurine Methyltransferase (abbreviated *TPMT*). This gene makes an enzyme that breaks down, or metabolizes, some medicines in the body. Breaking down a medicine can either make it work as intended or make it stop working. It's common to have slight variations in the *TPMT* gene that affect how the enzyme works to break down medicines called thiopurines. Depending on these variations, people are considered Poor, Intermediate, or Normal Metabolizers.

1. **Poor metabolizers** – The *TPMT* enzyme has very little activity. Poor metabolizers break down thiopurines very slowly so they stay in the body longer. Poor metabolizers are likely to need much lower doses of thiopurines than normal metabolizers or need different medicines. About one in 400 of our patients are poor metabolizers for *TPMT*.

2. **Intermediate metabolizers** – The TPMT enzyme has medium activity. Intermediate metabolizers need lower doses of thiopurines than normal metabolizers or need different medicines. About one in 10 of our patients are intermediate metabolizers for *TPMT*.
3. **Normal metabolizers** – The TPMT enzyme activity is normal. No thiopurine dose adjustments need to be made based on this result. About 9 out of 10 of our patients are normal metabolizers for *TPMT*.

## Your result was not able to be determined (uncertain).

This means that we are unable to predict the enzyme activity based on your result. Your doctor can use standard medication selection and dosing strategies that do not use genetic information. The following medicines are broken down by the TPMT enzyme:

**Thiopurines:** azathioprine, mercaptopurine, and thioguanine.

Research continues to be done on what medicines are affected by genetic test results. For more details about which medicines are broken down by *TPMT*, please go to [www.cincinnatichildrens.org/gps](http://www.cincinnatichildrens.org/gps) or [www.pharmgkb.org](http://www.pharmgkb.org).

If you have questions about your pharmacogenetic test results from CCHMC, call 513-636-4474 or email [gpsconsult@cchmc.org](mailto:gpsconsult@cchmc.org).

*Questions about individual health concerns or specific treatment options should be discussed with your physician.*

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