

## Patient information factsheet

# DPD deficiency and DPYD gene testing

We've given you this factsheet because we would like to offer you a genetic test before you start cancer treatment to check for dihydropyrimidine dehydrogenase (DPD) deficiency. It explains what DPD deficiency is, why we test for it and what the test involves. We hope it will help to answer some of the questions you may have. If you have any further questions, please speak to your oncologist (specialist cancer doctor).

### What is dihydropyrimidine dehydrogenase (DPD)?

DPD is an enzyme made by the liver. It helps break down certain chemotherapy drugs called fluoropyrimidines that can be used to treat cancer.

DPYD is a gene. Genes are our cells' instruction manuals. Our genes tell our cells how to function normally. The DPYD gene helps to control how much DPD enzyme is produced in the body. Everyone has two copies of this gene.

### What is DPD deficiency?

DPD deficiency is when a person has low (severe deficiency) or no (complete deficiency) levels of the DPD enzyme in their body. This is usually the result of a variant (change) in the DPYD gene.

A gene variant is inherited and can be passed on through your family. To have a severe or complete DPD deficiency, you need to inherit a variant in the DPYD gene from each parent. This is very rare and is usually diagnosed in childhood.

Having a DPD deficiency may mean your body doesn't have enough DPD enzyme to break down fluoropyrimidine chemotherapy drugs safely. This can make the side effects of fluoropyrimidines worse, even life-threatening for some people.

### Why am I being offered a DPYD gene test?

Approximately 2 to 8% of the population inherit a gene variant from one parent that reduces the amount of DPD they produce. However, there are no physical symptoms of DPD deficiency, so a DPYD gene test is the only way to find out if you have a deficiency. The results of this test will help your oncologist plan the best treatment for you.

### What does the DPYD gene test involve?

Before you start treatment for your cancer, your oncologist will arrange for you to have a blood test. This blood test will be sent to a genetic laboratory so that your DPYD gene can be examined. The result will be available in about five days and will be sent back to your oncologist.

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## What are the possible outcomes of testing?

There are three possible outcomes from DPYD gene testing:

### If both copies of your DPYD gene are working

You will be able to have a standard dose of fluoropyrimidine. This is because your body will be able to break down the chemotherapy drug.

### If only one copy of your DPYD gene is working and the other has a variant

You will be able to have fluoropyrimidines at a reduced dose. If your body tolerates the first treatment, your oncologist may be able to increase your dose.

### If both copies of your DPYD gene are not working

Your body is not able to produce the DPD enzyme. You will not be able to have fluoropyrimidines. Your oncologist will discuss alternative treatments with you.

Your oncologist will contact you to discuss your results and explain what the next steps are.

## What will happen if I decide not to have the DPYD gene test?

If you decide not to have the DPYD gene test, we will not be able to treat your cancer using fluoropyrimidines.

## What does a diagnosis of DPD deficiency mean for my family?

If your test results show you have a DPD deficiency, other members in your family may also have a DPD deficiency. This is because you share your genetics with blood relatives.

We do not offer testing for family members. This is because the chance of problems occurring for people with a DPD deficiency who are not receiving fluoropyrimidines is very small. However, we encourage you to share this information with your family in case they are treated for cancer in the future.

**Please be aware that your blood sample will only be tested for DPYD gene variants, so you will not find out about your risk of any other genetic conditions.**

## Contact us

If you have any questions or concerns, please contact your clinical nurse specialist.

## Useful links

Cancer Research UK: [www.cancerresearchuk.org](http://www.cancerresearchuk.org)

Macmillan Cancer Support: [www.macmillan.org.uk](http://www.macmillan.org.uk)

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