

# DPD deficiency and *DPYD* gene testing

Information for patients



Leeds Cancer  
Centre

You have been given this leaflet because your medical team are planning on treating you with a fluoropyrimidine chemotherapy drug, either 5-fluorouracil (5-FU) or Capecitabine.

It is known that people who have a low level of an enzyme called dihydropyrimidine dehydrogenase (DPD) are more likely to get severe side effects from these drugs. It is possible to find out who is most at risk of DPD deficiency by testing a blood sample for mutations (changes) in the *DPYD* gene. The safety of treatment can be improved if it is known who has a DPD deficiency before starting one of these drugs.

### What is DPD?

The enzyme DPD helps our bodies break down and process 5FU and Capecitabine. The *DPYD* gene provides instructions for making the enzyme DPD. If there are mutations in the *DPYD* gene the body will make less DPD enzyme.

### Why is it important to find out if I have DPD deficiency?

Without enough DPD, 5FU and capecitabine can build up in the body and cause more severe side-effects than usual. Sometimes these side effects can be life threatening.

It is very rare to have no DPD (a complete DPD deficiency). It is more common to have low or very low levels (a partial deficiency). Between two and eight out of every 100 people have a partial DPD deficiency.

Usually, we only become aware of DPD deficiency when side-effects occur whilst receiving chemotherapy with a fluoropyrimidine drug such as 5-FU or Capecitabine. This is why we test for *DPYD* gene mutations before giving these drugs.

If not tested for DPD deficiency before treatment starts, between 10 and 30 out of every 100 people have serious side effects when having treatment with 5FU or capecitabine. Around 1 in 200 people die from complications of their treatment. DPD deficiency is responsible for some of these side-effects.

By testing for the most common mutations in the *DPYD* gene that cause DPD deficiency we hope to reduce these serious side-effects.

### How do I get tested for DPD deficiency?

A small blood sample is taken and sent to the genetics laboratory. They look for certain mutations (changes) in the *DPYD* gene. The result is usually available within a week.

### What happens if I am found to have a *DPYD* gene mutation?

If the test finds *DPYD* gene mutation, then the importance of this will be discussed with you. Changes to your chemotherapy dose may be required to make your treatment safer. This will mean that your body should be able to process the drug and reduce it building up in your system and causing problems.

### What happens if I have a normal *DPYD* gene test result?

You will receive the planned dose of chemotherapy. It is however important to be aware that not all severe side-effects are caused by DPD deficiency or might be caused by a mutation that is not tested for. Some people still develop severe side effects even if they have a normal *DPYD* gene result. This means you may still get side effects as detailed on the specific drug treatment information. It is important that you read this information. You should report any side effects to your medical team without delay so you can get treatment quickly.

## What should I consider before having testing?

- The blood sample is only tested for *DPYD* gene mutations so you will not find out about your risk of any other genetic conditions.
- It is standard practice for the hospital to store your sample for 25 years in case there is a need to do future tests.
- Stored samples may be used anonymously for the development of new tests and quality checks.

Talk to your doctor or nurse if you are worried about *DPYD* gene testing and want any further information.

### Reference:

[www.cancerresearchuk.org/about-cancer/cancer-in-general/treatment/chemotherapy/side-effects/dpd-deficiency](http://www.cancerresearchuk.org/about-cancer/cancer-in-general/treatment/chemotherapy/side-effects/dpd-deficiency)

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