Cancer Council Victoria

FactSheet

Tumour testing for people at or under 50 years who are being treated for bowel cancer

Background
You have been diagnosed with bowel cancer and will be having an operation to treat it. You may already have had a biopsy to confirm the diagnosis.

We know that bowel cancer can, for some people, be partly due to changes in genes inherited from one of their parents. Even if neither parent had bowel cancer, it is possible that one parent carried a genetic change that increased their risk.

We believe that about one-third of people with bowel cancer have inherited a genetic risk for the disease. We have found the genes responsible for about 5% of bowel cancers. These four genes (called MLH1, MSH2, MSH6 and PMS2) are changed (or 'mutated') in this 5% of bowel cancers.

Because of your diagnosis at a young age (at or under 50 years) we would like to do some extra testing on your tumour while it is being examined by the pathologist. We would like to see whether there may be a change in one of these four genes in the tumour.

This is not a genetic test. However, it may show whether genetic testing may be helpful for you.

What tests will be done?
We do not require any tissue samples from you.

The tests will be done on tumour tissue taken in the routine course of your treatment (including biopsies and surgery).

When your bowel tumour is analysed by the pathologist, sections of it will be stained with a dye to highlight four proteins. These four proteins are made by the four genes mentioned above. If the genes are working normally they should be making all four proteins.

What is a normal result?
If staining is present in your bowel tumour for all four proteins it means that all of these genes are working normally. This means it is very unlikely that you have inherited a change (mutation) in one of these genes.

What is an abnormal result?
If there is no staining in your bowel tumour for one or more of these proteins it **MAY** mean that there is a change (mutation) in one of these genes.

What happens if you find an abnormal result?
Your treating doctor will discuss this with you and may suggest that you attend a Family Cancer Clinic. The reason for this is that you may carry a change (mutation) in a gene and you can have genetic testing to try to find this change.

An abnormal result may have implications for your treatment and for other family members. For example, it may suggest that you and/or family members could benefit from regular screening for bowel cancer.

What happens if you find a normal result?
If the result is normal it implies that all four of these genes are working normally and that you have no changes (mutations) in them.

However, there may be other types of genetic changes that could have affected your risk of bowel cancer.

If you have a family history of bowel cancer you should discuss this with your treating doctor even if this test is normal.

Will this test change my treatment?
The results are unlikely to change your treatment.