Southeast of Scotland Genetic Service
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Two Stage Genetic Testing for Lynch Syndrome

Information for Patients

Patient Label

Cancer is a common condition. It is not usually inherited. However, if we see more cancer in a family than we would expect to see by chance, if it starts at a very young age, or if certain patterns of cancer occur together, there may be an underlying predisposition to cancer running in that family caused by an inherited gene change. It is important to identify such families so that people who are at a higher risk of developing cancer than the general population can be offered appropriate screening or intervention.

We can sometimes detect such a change by genetic testing in someone who has had cancer. The testing is carried out in two stages. The **first stage** is to look at the colon tumour sample for specific features which are associated with an alteration in one of these genes.

- **If the tumour does not show these features**, then it reduces the likelihood that a known gene alteration is responsible and no further testing would be done.
- **If these features are present,** it may be just chance but it may also be that there is an underlying gene alteration. We would proceed to look at a sample of blood for alterations in the associated genes (**second stage**).

The genes associated with colon cancer predisposition are called the “mismatch repair” genes and people with a change in one of these genes are said to have a condition called “Lynch Syndrome”. In males the main cancer risk is in the colon and in women the main cancer risks are in the colon and also the uterus (endometrial cancer). There are other cancers associated with this condition but the risk of these is not as high. Other cancers include ovarian cancer and stomach cancer.

Possible outcomes of genetic testing

No gene change identified:
If testing in the tumour sample shows features suggestive of a genetic predisposition **but** an underlying gene change is **not** identified**,** we will assess your risk and your family members’ risk of cancers on the basis of the family history. We will not have a gene test to offer other family members. Occasionally we will proceed to test other genes if the family history indicates this may be useful.

“Pathogenic” or “Likely Pathogenic” (disease-causing) gene change identified:
If testing in the tumour sample shows features suggestive of a genetic predisposition **and** an underlying gene change is identified, this offers an explanation for your cancer development and may indicate you have an increased risk of developing another cancer. Identifying a gene change also means it is possible to offer a genetic test to other family members. This would increase the accuracy of their risk assessment and ensure screening and/or risk reducing strategies are appropriate.

“Variant that needs further assessment”:
We all carry variations in our genetic code. The majority of these are **not** linked to disease. If testing identifies a variant which requires further assessment, we will discuss this with you.

A limitation of any genetic test is that just like much of medical science, we do not know with certainty all the factors that influence health. Very occasionally the current understanding of a gene change or variant may change as more information becomes available.

Plan of action going forward

Please tick as appropriate		Enter details
	You have decided to proceed with testing and we have made the following arrangements for you to receive the result:	
	A follow up appointment has been arranged on:	
	You have decided to get back in touch when you feel ready to proceed with testing.	

We will write to your referring clinician/GP with details of the plan above.

Following testing, results are usually available within 3 months. Results are confirmed in writing to you and copied to your referring clinician/GP.

You were seen by:	Genetic Counsellor (contact number at top of first page)	Date: