Southeast of Scotland Genetic Service
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Combined Mutation Testing – Cancer Predisposition

Patient Label

Information for patients

Cancer is common and not usually due to an inherited cause. 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 genetic change.

It can be helpful to identify if a cancer is due to a genetic change because it may guide your cancer treatment and provide options to reduce your risk of future cancer. It can also provide information for relatives about their risks of cancer.

You have been offered genetic testing on the basis of your history of: _____

The testing will look for changes in the set (or panel) of genes currently known to be associated with this type of cancer.

Possible outcomes of genetic testing

No gene change identified:
In most people the test will not find a gene change. This reduces the chance of a predisposing gene being responsible for your cancer. However, other genetic factors could be involved if you have a family history of cancer. In these circumstances the genetics team will review your family history and offer advice for you to share with your family. There will not be an informative gene test available for other family members.

“Pathogenic” or “Likely Pathogenic” (disease-causing) gene change identified:
This confirms an inherited risk to cancer and is likely to explain your own diagnosis. Depending on the gene involved, it may mean that you have an increased risk of developing other type(s) of cancer. The genetics team will discuss what the test result means for your future risk of cancer, options for future screening, and measures to reduce these risks and address any questions you have. They will evaluate your family history and can provide information for appropriate family members should they wish to consider testing to see if they have inherited the gene change. Any relatives can be referred to their local Genetics Unit, to discuss this further. Your cancer team will use the information in their management decisions.

“Variant of uncertain significance (VUS)”:
We all carry variations in our genetic code, so it is not uncommon to detect a ‘variant’ when testing a number of genes. The majority of variants are not linked to disease. However, occasionally we do not have sufficient knowledge to know whether a change is significant in the development of cancer or just natural variation. We would not offer other family members a genetic test based on this result. You may wish to review this with us in the future.

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.

***The genes that you have agreed to be looked at have been ticked in the table below**		
Tick	**Type of Panel**	**Genes on Panel**
	Breast	*BRCA1, BRCA2, PALB2, PTEN, TP53, STK11, ATM, CHEK2, RAD51C, RAD51D*
	Ovarian	*BRCA1, BRCA2, BRIP1, MLH1, MSH2, MSH6, RAD51C, RAD51D*
	Breast and Ovarian	*BRCA1, BRCA2, PALB2, TP53 PTEN, STK11, ATM, CHEK2, BRIP1, MLH1, MSH2, MSH6, RAD51C, RAD51D*
	Colorectal/ Lynch	*APC, BMPR1A, MutYH, PTEN, SMAD4, STK11, MLH1, MSH2, MSH6, PMS2, POLE(exons 3-13) POLD1(exons 4-12), RNF43, NTHL1, MBD4, GREM1 (upstream duplication testing for polyposis referrals only)*
	Prostate	*BRCA1, BRCA2, CHEK2, ATM, PALB2, TP53, MLH1, MSH2, MSH6, RAD51D, PMS2, EPCAM, HOXB13*
	Pancreatic	*BRCA2, CDK4, CDKN2A, MLH1, MSH2, MSH6, PALB2, STK11, TP53*
	Skin	*BRCA2, CDK4, CDKN2A, POT1, BAP1*

Plan of action going forward

Please tick as appropriate		**Enter details**
	You have decided to proceed with testing. We have made the following arrangements for you to receive the results:	
	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.

Results will be 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: