Southeast of Scotland Genetic Service
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Genetic testing of stored sample from deceased cancer patients

Living person's Patient Label

Information for Relatives

Your deceased relative may have a sample stored in the laboratory. We can request testing of this sample for changes in genes* known to be associated with cancer risk.

Cancer is common and is 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 was due to a genetic change because it can provide information for relatives about their risks of cancer, guide options to reduce cancer risk or guide treatment options.

The testing of your relative's sample 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

Genetic testing not possible:
Testing may not be possible if there was no sample taken or stored. Alternatively, samples may have been stored in a preservative that causes genetic testing to not be possible. In these circumstances the genetics team will review your family history and offer advice. There will **not** be an informative gene test available for family members.

No gene change identified:
In most people the test of your relative's sample will **not** find a gene change. This reduces the chance of a predisposing gene being responsible for your relative's 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 that your relative had an inherited risk of cancer. A genetic test would be available to relatives to clarify their risk. The genetics team will discuss this with you and address any questions you have. They will discuss what the test result means for your future risk of cancer, your options for future screening, and measures to reduce these risks. They will evaluate your family history and can provide information for the 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.

"Variant of uncertain significance (VUS)":
We all carry variations in our genetic code. The majority of these 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.

Plan of action going forward

Please tick as appropriate		Enter details
	You have decided to proceed with testing of your deceased relative's sample. 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 of your deceased relative's sample.	

We will write to your referring clinician/GP with details of the plan above. Results are confirmed in writing to you and copied to your referring clinician/GP.

Seen by	Genetic Counsellor (contact number at top of first page)	Date:

***The genes to be examined have been ticked in the table below**		
	Breast	*BRCA1, BRCA2, PALB2, PTEN, TP53, STK11, ATM, CHEK2, RAD51C, RAD51D*
	Ovarian	*BRCA1, BRCA2, BRIP1, MLH1, MSH2, MSH6, RAD51C, RAD51.*
	Breast & 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, ATM, HOXB13 – standard panel.* *BRCA1, BRCA2, CHEK2, ATM, TP53, MLH1, MSH2, MSH6, RAD51D, PMS2, EPCAM, HOXB13.* ***Inc PALB2** (only if family history of breast cancer)*
	Pancreatic	*BRCA2, CDK4, CDKN2A, MLH1, MSH2, MSH6, PALB2, STK11, TP53*
	Familial Melanoma	*BRCA2, CDK4, CDKN2A, POT1, BAP1*