Southeast of Scotland Genetic Service
Western General Hospital, Crewe Road, Edinburgh, EH4 2XU
Tel: 0131 537 1116

Patient Label

Genetic Testing for Multiple Colon Polyps

Information for Patients

Because of your diagnosis of multiple colon polyps, you are being offered a test to look for changes in the genes* currently known to be associated with an increased risk of developing colon polyps.

Polyps can occur in an individual with no family history and not due to a genetic cause. Some families, however, have an increased risk of developing multiple polyps due to an underlying genetic predisposition that is inherited through the family.

There are a number of known genetic conditions that predispose to polyp development and have an associated risk of developing colon cancer.

We can sometimes detect such a change by genetic testing using a blood (or saliva) sample from someone who has developed multiple polyps. This can be helpful to guide management and provide options to reduce risk of future cancer. It can also provide information for relatives about their risks of developing colon polyps and cancer.

There are 3 possible outcomes of genetic testing:

- **No gene change identified:**

This reduces the probability of a genetic cause but does not completely rule it out. Your consultant surgeon will make a decision on how frequently colonoscopies should be completed. We will assess your family members' risk on the basis of the family history and offer advice on screening measures, where appropriate. There will not be a genetic test to offer them.

- **"Pathogenic" or "Likely Pathogenic" (disease-causing) gene change identified:**

This offers an explanation for your polyp development and indicates that you have an increased risk of developing further polyps, and possibly colon cancer. Depending on which gene the change is in, there may be an increased risk of developing other types of cancer. Identifying a gene change also means it is possible to offer a genetic test to your 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 material (DNA). 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 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.

*Current Panel: APC, BMPR1A, MUTYH, PTEN, SMAD4, STK11, MLH1, MSH2, PMS2, NTHL1, POLE, POLD1, GREM1 (upstream dup)

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 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.

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: