

RELATIVE INFORMATION

Whole genome sequencing study of young colon cancer patients and their parents



Colorectal cancer (bowel cancer) is a major health problem in the United Kingdom. It is important that we try to improve our understanding of why it occurs in young people, with the help of their relatives, so that new ways of preventing and treating bowel cancer can be developed.

We would like to invite you to take part in a research study. Before you decide it is important that you understand why the research is being done and what it would involve for you. Please take time to read the following information carefully. Talk to others about the study if you wish. Please ask if there is anything that is not clear or if you would like more information.

Take time to decide whether or not you wish to take part and discuss the study with your family.

What is the purpose of the study?

Whilst cancer of the large bowel is common, it is very rare in young people under the age of 40 years. Our aim in this study is to look at samples (blood and tissue removed from tumours) from individuals who have developed bowel cancer aged 40 years or younger and also blood samples from each of their unaffected parents. These samples will be analysed using a technique called "Next-Generation Sequencing" (NGS).

NGS is a scientific technique that gives us a "read-out" of all the genetic information that is stored in our DNA within each of our cells within the body. It is this information that makes every person unique. In doing this, and by comparing the sample from your son or daughter, we aim to identify changes in their DNA (mutations) which may not present in their parents. We aim to collect this information to help us identify mutations that are causing bowel cancer. This will, in the long term, help us to develop new treatments and predict who will be susceptible to cancer and so be able to prevent disease progression in the general population.

Why have I been invited to take part?

You have been asked to take part because you are eligible as the parent of a child who has previously had a diagnosis of a bowel cancer before the age of 40 years who does not have an underlying genetic cause or any close family members affected by the disease. With your help, we would like to try and understand why someone in your family has developed bowel cancer so young when there is no evidence of bowel cancer in their parents or grandparents. It is important that both parents are able to provide a blood sample.

Do I have to take part?

You do not have to take part. It is up to you and other family members to decide. We will describe the study and go through this information sheet with you. Your son or daughter will also have the chance to talk through the study with you. If you are all willing to take part you can contact the study team directly, or inform a member of your healthcare team who will let us know. You will be asked to sign a consent form to show that you understand the risks and benefits of the study.

Do I have to provide a blood sample?

Blood is the most valuable biological material for analysing DNA and we would prefer to conduct our studies on blood DNA. However, a saliva sample may be provided from individuals unwilling or unable to provide a blood sample.

What will happen to me if I take part?

If you decide to take part, it should take no longer than 15 minutes. This can be done by accompanying your son or daughter to a routine clinic visit or at a time that is preferable to you. You will be asked to:

- Give a one-off blood sample of approximately 6 teaspoons (30mls) for next generation sequencing and blood biomarkers or a simple saliva sample.
- Give information about your family history by answering some brief questions about your relatives and whether any of them have had bowel cancer.
- Give some very basic information about your health and medication history.
- Give permission for access to your medical records for any study specific information during the course of the research.
- Give permission for a small amount of personal information to be sent to the research coordinating centre in Edinburgh.
- Give permission for your sample to be stored for future research studies.
- What are the possible risks and benefits of taking part?



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The blood sample would be taken through a needle, usually in your arm, which can sometimes cause slight bruising. We can try to take this at the same time as you are having any other blood samples done if you are currently under the care of your General Practitioner. There is limited risk in providing a saliva sample.

It is possible that this study will not directly benefit you. However, should anything be identified during the course of the research which may be important to your clinical care now or in the future, or may be of importance to your family, we would inform your healthcare team of the results and this will be discussed with you.

Will the research have any implications for my health?

The studies that we will conduct using your sample and data will allow us to understand the causes of bowel cancer in a younger population. It may not generate information that would be individually useful to any future clinical care or that of your family. Nonetheless, if the research were to reveal results which may have relevance to you or your family, they will contact your healthcare team directly who will notify you of the results. However, it may take a considerable period of time and much follow up work to show that any genetic changes that are identified, are in fact causing cancer, rather than merely being present in individuals with bowel cancer. As such, any results that could be of use to you or family may take some time.

Will my taking part be kept confidential?

Yes. We will follow ethical and legal practice and all information about you will be handled in strictest confidence. Your blood sample will be coded and have no identifying information on it. Only members of the University of Edinburgh research team will have access to the named data. Data on computer will be accessible only by research team members and will be password protected and stored within a highly secure computer system in a secure building. Any written information will similarly be stored securely in locked premises.

What will happen to the results of the study?

The overall results of the project will be made widely available. They will be published in the medical literature but always in an anonymous way so results cannot be traced back to individual people.

Who will have access to samples?

As well as the research team in Edinburgh, we work in collaboration with research teams from the UK and abroad to test theories about the causes of bowel cancer. We will sometimes use your samples and anonymised information for these.

In all cases only projects passed by research ethics committees and international research review boards will be approved. In the future such sample collections may be useful to commercial companies in the development of treatments. These applications will be subject to the same close scrutiny and ethical review.

What happens if I change my mind?

You can withdraw from the study at any time without giving a reason. This will not affect any health care you or your relatives receive. Your samples and any other information will be destroyed. If you wish to withdraw from the study, please contact the study office at the address below.

Who is funding the research?

The research is funded by Cancer Research UK, the largest cancer charity in the UK.

Who has reviewed the study?

All research in the NHS is looked at by an independent group of people called a Research Ethics Committee to protect your safety, rights, wellbeing and dignity. This study has reviewed and given a favourable opinion by Regional Ethics Committees throughout the United Kingdom and regional National Health Service sites.

What will happen now?

You will be asked to discuss the study with your family and decide if you wish to take part. If you are willing to participate, or have any questions, please do contact the study office directly. They will be able to answer any queries you may have and help make arrangements with your healthcare team for your participation.

Thank you for reading this information sheet.

If you wish to speak to someone not involved in the study, or have a complaint, you can contact:

Professor David FitzPatrick, Senior Clinical Scientist, MRC Institute of Genetics & Molecular Medicine, The University of Edinburgh, Western General Hospital, Crewe Road, Edinburgh EH4 2XU

Telephone 0131 651 8569

If you have any queries about the study please contact:

The SCOTTY Study, MRC Human Genetics Unit, MRC Institute of Genetics & Molecular Medicine, The University of Edinburgh, Western General Hospital, Crewe Road, Edinburgh EH4 2XU

Study office telephone/answer service
0131 651 8550 or 0131 651 8602

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