PATIENT INFORMATION

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

the SCOTTY study
SEQUENCING OF COLON TRIOS IN THE YOUNG

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 so that new ways of preventing and treating bowel cancer can be developed in the general population.

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. Your parents will also be asked to consider the study. 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 parents.

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. Our aim in this study is to look at samples (blood and tissue removed from tumours) from individuals who have developed bowel cancer at a young age and also blood samples from each of their 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 comparing your sample to those of your parents, we aim to identify changes in your DNA (mutations) that are not present in your 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.

Why have I been asked to take part?

You have been asked to take part because you have 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. It is important that both your parents are able to provide a blood (or saliva) sample and they will also be asked to consider the study.

Do I have to take part?

You do not have to take part. It is up to you and both your parents to decide. We will describe the study and go through this information sheet with you and with your parents. If you are all willing to take part you can contact us directly or inform a member of your healthcare team who will let us know. You and your parents 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 next generation sequencing 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 during a routine clinic visit or at a time that is preferable to both you and your parents. You will be asked to:

- Give a blood sample of approximately 6 teaspoons (30mls) for next generation sequencing (NGS) and blood biomarkers or a saliva sample.
- Give permission to access any stored DNA from the Regional Clinical Genetic Centre.
- Give permission to access archived stored tumour samples from the time of your operation.
- Give information about your family history by answering some brief questions about your relatives and whether any of them have had bowel cancer.
- Give information about your treatment, your medical and medication history.
- Give some basic measurements including your height, weight.
- Give permission for access to your medical records (both your medical notes and any electronic medical records) for study specific information during the course of this research. This will enable us to follow your treatment without asking you each time. Access to social care records will not be required.
- Give permission for a limited amount of personal information to be transferred to the research coordinating centre in Edinburgh.
- Give permission for your sample to be stored for future studies.

What are the possible risks and benefits of taking part?

The blood sample would be taken through a needle, usually in your arm, which can sometimes cause slight bruising. We will try to take this at the same time as you are having any other blood samples done. There is limited risk involved in supplying 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, or may be of importance to your family, we would let your healthcare team know. We may advise your healthcare provider that it would be of benefit to you to be referred to your local Genetics Unit for further support and counselling if they have not already done so.

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 produce information that would be individually useful to your clinical care or that of your family. Nonetheless, if the research were to reveal results that has relevance to your medical condition, or to your family, the research team will contact your healthcare team 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. The sample you provide will be coded and have no personal identifying information on it. Only approved members of the University of Edinburgh research team will have access to your 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 sample 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 Committee's throughout the United Kingdom and regional National Health Service sites.

What will happen now?

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