Division of Genetics
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Participant Information Sheet V3 **25/10/2012**

Study Title: Targeted Recall of UK10K Cohort – Cambridge BioResource
Recruitment of healthy volunteers to understand the function of immune pathways predisposing to autoimmune disease

Invitation: You are being invited to take part in a research study, which will help scientists to understand more about how the immune system works. Before you make your decision to participate it is important for you to understand why we want to do the study and what you will have to do to take part. Please take time to read the following information carefully and discuss it with others if you wish. If after reading all the information, you still have any more questions, then please contact me at:

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Please take time to decide whether or not you wish to take part. Thank you for reading this information sheet.

A) What is the scientific purpose of the study?

We are studying the autoimmune disease Systemic Lupus Erythematosus (SLE), which affects mainly young people and can cause a range of problems including skin rashes, arthritis, kidney disease and brain disease. Although the cause of this disease is so far unknown, we know that genes (currently about 50 have been identified) are important in this process. Understanding how these genes control the cells and proteins of the immune system will give us important information about where to look to understand further what is happening in patients with disease, and therefore which parts of the immune system could be targeted with new treatments. We have found that trying to conduct these studies in patients with SLE is difficult, firstly because the drugs patients take alter the components of the immune system we are looking at and, secondly, because the disease itself causes a lot of changes to the immune system which are a consequence of the disease but not part of the cause of it. Since most of the gene variants we are interested in can also be found in healthy people we think that trying to understand how these networks of genes work in people without disease will be much more productive.

B) How can I participate and what does it involve?

1) ***Why have you been chosen?*** You have been chosen based on your initial recruitment into the Cambridge BioResource and on the basis of your genetic make-up determined from your first donated blood/saliva sample.

2) ***How can you help?*** We are inviting volunteers to participate who are prepared to provide up to a 60ml sample of blood and travel to Cambridge for donation of the sample (the visit will be short and reasonable travel costs will be re-imbursed). The blood samples will be used to study how genes influence the risk of disease development.

The purpose of the study is simply to look at how different changes in genes affect the way that they do their job within the body. We want to collect blood samples from a large number of different people; it is helpful to us to look at genes with a lot of different combinations of changes (from lots of different participants in the study). In this way it will be easier for us to work out whether any of those changes (or combinations of changes) alters the way that a gene behaves in your body.

So you may ask," If I have lots of changes in my DNA, why don't I have lots of diseases?" This is because most common diseases are caused by a collection of many disease-causing changes in addition to other factors in our environment, such as infection, stress or fatigue. Each single change will not be strong enough to cause a disease on its own, but may change the way that some of the genes work in our bodies. It is the combination of lots of changes in lots of genes which will cause disease. However, it is important to remember that most changes in our DNA are probably "silent", that is, they don't cause disease.

C) Practical details in taking part in the study

1) **Having been contacted, do I have to take part?** You are under no pressure at all to take part in the study and you should not take part in the study if you do not wish to do so. Furthermore, if you decide to take part you are still free to withdraw at any time and without giving a reason (**see Section E for further details**).

2) **What will happen to me if I do want take part?**

STEP 1: Your decision to take part

Whether you would like to take part in the study or not we ask that you complete and return the participation slip sent with the invitation letter you received to let the CBR team know your decision.

If you do decide to take part in the study you will be contacted by telephone and invited to take part in this study. We will not be able to include any participants in the study who are on long term medication that may impact on the function of the immune system, who have an autoimmune disease or who are pregnant.

STEP 2: At your visit

On arrival at the Cambridge BioResource you will be met by the research coordinator who will explain the study to you and answer any questions that you may have. Once you have agreed to continue you will be asked to sign the consent form.

STEP 3: Giving the blood sample

We will use the blood sample to prepare blood cells to investigate how genes work.

3) **What will happen to my blood samples and the information that we gather on them?**

Your blood sample will be taken and will be labelled with a unique code identifier only.

Secure storage of your personal information

None of the research scientists working on your sample will be able to identify you from the coded label.

Secure storage of your blood **samples** Once we have received your blood sample, it will be stored in a secure freezer. During the course of the experiments and subsequent investigations, your sample will only ever be referred to by its unique code identifier.

Secure storage of the experimental results on your sample All of the experimental information gathered on your sample will be stored under the unique code identifier, in the Dept of Genetics and Molecular Medicine. The only people who will have direct access to this information will be the scientists directly involved in the project.

D) Other Information

1) What will happen to the results of the research study?

The data/intellectual property collected as a result of your participation will always reside with the Hospital and Kings College London.

The results of the genetic analyses will be published in scientific journals. Due to the nature of the work it will be difficult for non-experts to understand all the published material since this is written in technical language and is aimed at other scientists. We will undertake to provide more generally understandable summaries of the research if requested.

Publications Published results from any papers using your data will not mention you by name and your individual results will not be traceable through any published material.

2) Will I receive any feedback on my sample?

We will not be able to supply you with your own individual genetic results or biological information from this research.

3) Possible advantages and disadvantages

What are the possible benefits of taking part?

There will be no direct benefits to you personally. Your participation will increase our understanding of immune-related diseases, which will hopefully allow the design of better and more specific treatments for autoimmune and allergic diseases.

What are the possible disadvantages and risks in taking part?

Some discomfort may occur when the needle is placed in the vein and the blood is drawn and there is the possibility that a bruise may develop. Light-headedness and fainting may also result from blood sampling. You should not take part if you know that you are pregnant, as it is better to avoid unnecessary blood sampling during pregnancy.

If you have any concerns about any aspect of the way you have been treated during the course of this study then you should immediately inform the Investigator (Professor Tim Vyse).The normal National Health Service complaints mechanisms are also available to you.

4) **Who is organising and funding the research?** The bulk of funding for this research comes from Arthritis Research UK.

5) **Who has reviewed this study?** The project has been subject for ethical review by South East London Research Ethics Committee 2.

E) Withdrawing from the study

A decision to withdraw at any time, or a decision not to take part, will not affect the standard of care you receive. If you do want to withdraw, you can contact Dr. Ben Rhodes, Clinical Senior Lecturer on Tel: 020 7188 3705 or write to him at the Dept of Genetics and Molecular Medicine, 7th Floor Tower Wing, Guy's' Hospital Campus, Great Maze Pond, London SE1 9RT.

You will not have claim to either the material that we collected or the result arising from the study. All results are anonymised and cannot lead to the identification of a single individual.

F) Contacting us for further information.

You are welcome to show this sheet to your family, friends and general practitioner if you would like to discuss any aspect of the study with them - and we would also be pleased to answer any questions that you may have. If you would like further information or to discuss the project further, please feel free to contact either of us by phone or email.
Please retain a copy of the information sheet and one copy of the signed consent form.

Thank you very much for your help.

Prof Tim Vyse