UNIVERSITY OF CAMBRIDGE
Autism Research Centre

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Participant Information Sheet A

Study Title: A Genetic Study of Autism Spectrum Conditions and Related Traits

This sheet will explain why this research is being done and what your participation would involve. Please take time to read the following information carefully.

What is the purpose of the study?

Genetic variations have been identified to cause Autism Spectrum Conditions (ASC). The aim of the study is to detect genetic variations in the human genome that cause ASC and related conditions. This study will include individuals with and without a diagnosis of ASC.

Why have I been invited?

We are contacting you because you are registered at the Autism Research Centre (https://autismresearchcentre.com) and indicated that you are diagnosed with ASC or have a family member diagnosed with ASC.

Do I have to take part?

It is up to you to decide. The study procedure is described in this information sheet, and if you agree to take part you will need to sign the consent form sent to you. You are free to withdraw at any time, without giving a reason. This will not affect the standard of care you receive.

What will happen to me if I take part?

Step 1: If you decide to take part in the study, please login here https://dnastudy.autismresearchcentre.net and indicate your agreement to receive a consent form and a saliva kit as well as to update your current postal address. If you have any questions, please feel free to email Clara Buckingham at cb905@medschl.cam.ac.uk.

Step 2: We will send you the consent form and a DNA saliva kit for collection of genetic information. Please complete both and send back to us using the provided envelope. In addition, if you are diagnosed with ASC, please provide us with a copy of a clinical report with information regarding your ASC diagnosis.

To use the saliva kit all you will need to do is spit into the tube. It is important that you **do not eat/drink/chew gum/smoke or brush your teeth for at least half an hour before providing a saliva sample** as these actions may contaminate your saliva sample, rendering it unusable.

Step 3: Log in to https://autismresearchcentre.net and complete the following questionnaires online, if you have not already done so: 'Your personality', 'Your feelings' and 'Your interests'. Feel free to complete additional tests and questionnaires once you have completed these.

Your anonymised DNA sample will be compared to DNA from other individuals to detect genetic and genetic-related differences that may contribute to the development of ASC or related conditions. If you have already taken the tests for previous studies at the Autism Research Centre,

(See more overleaf)

we ask for your permission to view the results. The DNA samples will be analysed by a member of our research team or an academic collaborator.

What are the possible disadvantages and risks of taking part?
We may share your anonymized (genetic and questionnaire) information in highly secure research databases or share with potential collaborators for future research. You can indicate if you agree for us to deposit your anonymized genomic data in research repositories in the consent form.

What are the possible benefits of taking part?
There are no direct benefits; however, the information we gain from this study will help to further our understanding of the relationship between genes and ASC.

What happens at the end of the study?
Results will be presented at scientific/medical research conferences and written up in journals. The data will be totally anonymous, without any means of identifying the individuals involved.

Who is organising and funding the research? Will I be paid?
This study is being organised by the University of Cambridge and funded by the Autism Research Trust. Unfortunately, we are unable to pay you for your participation in the study.

Will my taking part in the study be kept confidential?
Yes. Your information will be stored without identifying details and will only be marked with a code, thus preserving your anonymity.

What will my DNA be used for?
We hope to use your DNA to search for structural and functional changes in the DNA that may contribute to ASC and related conditions. This will be done by analysing either your entire DNA code or only parts of your DNA code. We may sequence all or parts of your DNA code, or search for genetic markers using a process called genotyping or related methods. We may also investigate how your genes are switched on or switched off, a process known as epigenetics. We are happy to describe this in detail should you need more information on this.

Will the research team identify clinically significant information in my DNA?
No. We are not directly testing to identify clinically significant genetic changes. Our genetic analysis is for research purposes only and is not for clinical diagnosis.

What will happen if I don't want to carry on with the study?
If you wish to withdraw from the study, you are free to do so at any point ewithout giving us a reason. If you wish us to destroy your DNA sample, we will do so upon request.

Who has reviewed the study?
This study has been reviewed and approved by an independent group of people at the Cambridge Psychology Research Ethics Committee.

Problems and further details:
If you have any concern or need further details, please contact Clara Buckingham (cb905@medschl.cam.ac.uk Tel: 01223 465226). If you are happy to participate, please email Clara Buckingham with your postal address and telephone number.

If you do not wish to participate, then you do not have to do anything. We hope that you will be interested in this study and will choose to participate in this project.