



Participant Information Sheet

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 have seen the study advertised in the press or on social media, and have indicated that you are 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 register at <https://dnastudy.autismresearchcentre.net> to indicate your agreement to receive a consent form and a DNA saliva kit. You will then be asked to answer some questions about yourself and then answer three questionnaires: 'Your Personality', 'Your Feelings' and 'Your Interests'. This should take no longer than 15-30 minutes. If you have any questions, please feel free to email Paula Smith at research@autismresearchcentre.com.

Step 2: We will send you a study kit consisting of the consent form, a return slip and a DNA saliva kit for collection of genetic information. Please complete all of these and send them back to us using the provided pre-paid 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 (the page that confirms the diagnosis) in the pre-paid envelope.

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: At the end of the online registration process you will be given the option to receive emails about other studies being conducted by the Autism Research Centre. If you are interested in helping with other studies please tick the box to be placed on our mailing list. You will receive a maximum of one email per month and are free to withdraw from the mailing list at any time by emailing Paula Smith at research@autismresearchcentre.com. You are under no obligation to take part in any of these studies.

Step 4: 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.

The DNA samples will be analysed by a member of our research team or an academic collaborator.

Step 5: 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 included in your study kit.

What are the possible disadvantages and risks of taking part?

- 1. Clinical reports via email/post:** There is a small risk that if you have a diagnosis and send us a copy of your clinical report, this could get lost in the post. Our freepost envelopes have worked very well, but if you are worried about this, we can arrange for yours to be sent by registered post (more secure) or a courier.
- 2. Potential identifiability:** Your DNA will be anonymised so there is no risk that anyone could identify you from your DNA sample.
- 3. Not receiving DNA data (health or ancestral information):** We regret that we cannot provide information about your genetic make-up back to individuals, as we only report on group results.

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 and the Templeton World Foundation, Inc. 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 without 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.

Cambridge University is the sponsor for this study based in the United Kingdom. We will be using information from you and/or your medical records in order to undertake this study and will act as the data controller for this. Cambridge University will keep identifiable information about you for 10 years after the study has finished.

Your rights to access, change or move your information are limited, as we need to manage your information in specific ways in order for the research to be reliable and accurate. If you withdraw from the study, we will keep the information about you that we have already obtained. To safeguard your rights, we will use the minimum personally-identifiable information possible.

You can find out more about how we use your information at

<https://www.medschl.cam.ac.uk/research/privacy-notice-how-we-use-your-research-data/>

Problems and further details:

If you have any concern or need further details, please contact Paula Smith at research@autismresearchcentre.com. If you are happy to participate, please complete the online registration process.

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.