Principal Investigator
Simon Baron-Cohen
Director, Autism Research Centre
Douglas House
18b Trumpington Road
Cambridge CB2 8AH

Participant Information Sheet
(Welfare Attorney/Welfare Guardian/Nearest Relative)
Study Title: Spectrum 10K

REC Ref: 20/SS/0080
IRAS Ref: 277521

You are being invited to consider giving your permission for your ward/relative/person you are consenting for to take part in a research study. Before you decide it is important for you to understand why the research is being done and what it will involve.

We will then ask that you put your own views about the research aside and to consider and take into account, the past and present wishes and feelings of your ward/relative/person you are consenting for, had they been able to consent for themselves.

Please take time to read the following information carefully and discuss it with others if you wish.

Please ask if there is anything that is not clear or if you would like more information. Thank you for reading this.

1. About Spectrum 10K

1. 1 What is the purpose of the study?

Spectrum 10K aims to recruit at least 10,000 individuals with a diagnosis of Autism Spectrum Conditions (hereafter 'autism') and, where possible, their relatives to identify genetic and environmental factors that contribute to autism and related conditions.

Spectrum 10K is funded by the Wellcome Trust and is being led by a team of researchers at the Autism Research Centre at the University of Cambridge, the Wellcome Sanger Institute and the University of California Los Angeles (UCLA).

Signing up to Spectrum 10K (at www.spectrum10k.org) will involve giving consent on behalf of your ward/relative/person you are consenting for to take part, completing questionnaires, providing their saliva samples, and giving consent to obtain their medical records (to investigate important clinical information relevant to the study). Participation in this study is completely voluntary. You can withdraw your ward/relative/person you are consenting for at any time without giving a reason.

2. Taking Part in Spectrum 10K

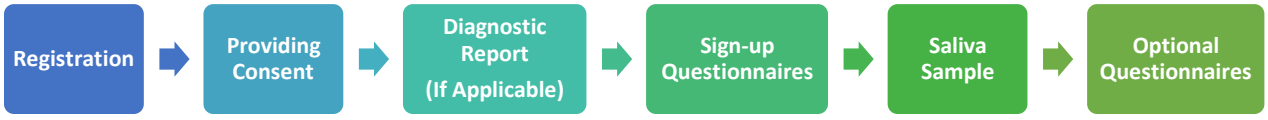
2.1 Why has your ward/relative/person you are consenting for been chosen?
Your ward/relative/person you are consenting for has been asked to take part because you have seen the study advertised in the press and/or on social media, or have been invited by their relative, or have been approached by their clinician, and you have indicated that your ward/relative/ person you are consenting for:

- Is aged 16+ and has been diagnosed with autism.
  OR
- Is aged 16+ and is biologically related to an autistic adult or child who is participating in Spectrum 10K (up to third-degree relatives which include: parent, sibling, child, aunt, uncle, niece, nephew, grandparent, grandchild, half-sibling, first cousin, great-grandparents).

However, they currently lack the capacity to make an informed decision about whether they can take part in a research study. We are therefore asking you as their Welfare Attorney/Welfare Guardian/Nearest Relative, if you will give consent on their behalf to join this study. This is permissible under the Adults with Incapacity (Scotland) Act 2000.

2.2 Do they have to take part?
No. It is up to you to decide whether they take part in the research or not. If you decide that they should take part you are free to change your mind at any time and without giving a reason and this will not alter their care in any way, now or at any stage in the future.

2.3 What will happen to your ward/relative/person you are consenting for if they take part in the research?



Participation in Spectrum 10K will not form part of standard care/ hospital /clinic appointments. Participation can take place in your own home as long as you have access to a computer. If you have been approached through a local autism clinic, a member of their team may help provide support to complete the study steps.

Step 1- Registration: If you decide your ward/relative/person you are consenting for would be happy to participate in the study, please register at www.spectrum10K.org. You will be asked to complete the following:

- **Consent:** You will be asked to sign a Welfare Attorney/Welfare Guardian/Nearest Relative consent form and consent on behalf your ward/relative/person you are consenting for. Once signed, the consent form will be available to download for your records.

  Some of the items on the consent form are optional. If you decide to opt out of any optional items, your ward/relative/person you are consenting for will not be excluded from the study, however all other consent points are required for participation.

- **Participant Information:** You will be asked to provide the name, date of birth, information about their autism diagnosis (if applicable) and demographic information of your ward/relative/person you are consenting for. We will also ask for the contact email address and the address to send the saliva kits to (if needed) for your ward/relative/person you are consenting for. This may be your address.

  We will also ask you to provide, if possible, the NHS number of your ward/relative/person you are consenting for to link to their Electronic Health Records. Please see below for more information about collecting information about their medical records.

- **Clinical Report**: If your ward/relative/person you are consenting for has an autism diagnosis from a qualified professional, you will be asked to upload a copy of their clinical report with information about their diagnosis (only the page that confirms the diagnosis). If you do not have an electronic copy, you may upload a photographed or scanned copy, as long as it is legible. This report will be uploaded into a secure database that can only be accessed internally by approved Spectrum 10K research personnel.

  Alternatively, you can provide a paper copy of their clinical report. We will provide you with a pre-paid return envelope if you choose this option.

  This is not mandatory and will just enable us to obtain more information about their autism diagnosis (if applicable).

- **Sign-up Questionnaire (Mandatory Baseline Questionnaire):** You will be asked to complete a sign-up questionnaire on behalf of your ward/relative/person you are consenting for. This includes questions about their health, lifestyle, education and questions related to autistic traits. This will need to be completed even if the individual you are consenting for is not autistic. This usually takes 15-20 minutes.

- **Coronavirus Questionnaire (Optional):** We want to understand the impact of COVID-19 on the autism community. At baseline, you will be provided with the option to complete an additional COVID -19 questionnaire on behalf of the individual you are consenting for. **This is optional** and will not stop the individual you are consenting for participating in Spectrum 10K if it is not completed. The questionnaire will ask about whether the individual you are consenting for was unwell due to COVID-19 or knew a person who was, symptoms they experienced, treatment management and shielding. It will also ask about the impact of COVID-19 on their finances, mood and anxiety levels. If you choose to complete the COVID-19 questionnaire on behalf of the individual you are consenting for, we will ask you to repeat the questionnaire every 3 months for 2 years. You will receive an email reminding you to complete the questionnaire when it is due.

Step 2 - Saliva DNA Kits: We will send a spit tube to provide your ward/relative/person's saliva sample, instructions on how to do so will be included along with pre-paid return envelopes. This will be sent to the address you provide. We will extract DNA from their sample. If you are at an NHS Trust site when registering your ward/relative/person, the research care team will be able to help you collect their saliva sample.

To use the saliva kit all your ward/relative/person you are consenting for will need to do is spit into the tube. <u>If this might be difficult to complete, we have assisted saliva collection kits. You will have the option to request the assisted saliva collection kit during registration.</u> If you are

at an NHS Trust site when completing this, the research care team will be able to provide you with an assisted kit.

Step 3 – Additional Questionnaires (Optional): You will have the option to log into your Spectrum 10K account (at www.spectrum10k.org) and complete additional questionnaires on behalf of your ward/relative/person you are consenting for. There are 3 sets of additional questionnaires for participants without capacity. **These are optional**. You can complete these all at once, or save your progress and complete these in your own time. We will send you a maximum of 4 reminder emails for each set of questionnaires over a period of 2 years after you register.

Additional optional questionnaires will include sensitive questions related to the mental and physical health of the person you are advising for and include potentially distressing topics such as abuse and suicide which you or the person you are advising for may find upsetting. If you decide to complete these questionnaires, you will have the option to stop at any time, refuse to answer individual questions, or skip entire questionnaires altogether. You may also wish to consider who is present when you are completing these to ensure that you are comfortable doing so.

2.4 Can other family members participate in the Spectrum 10K study?
Yes, if your ward/relative/person you are consenting for has a professional autism diagnosis, their biological relatives can be invited to participate in Spectrum 10K (up to third-degree). Third degree eligible relatives include: biological parents, biological children, biological (half or full) siblings, biological aunts or uncles, grandparents, first cousins, and great-grandparents.

If the ward/relative/person you are consenting for does not have an autism diagnosis, then you will not be eligible to invite these relatives.

If eligible, you will need to specify the relationship of your ward/relative/person you are consenting for to each individual and enter their email address. Relatives will receive a URL link which will direct them to register online. Using this URL method will allow for relatives to be automatically linked to your ward/relative/person you are consenting for. We will not collect any personal information about relatives at this point and the email address will not be visible to us.

Relatives who are invited to participate in the study must complete their own online registration form, including consent and will be asked to provide their own saliva sample. All child relatives must be registered by a parent or legal guardian.

2.5 Will I or my ward/relative/person I am consenting for be paid?
Unfortunately, we are not able to pay you or your ward/relative/person for your or their participation in this study.

2.6 Will I or my ward/relative/person be re-contacted?
To better understand co-occurring conditions and wellbeing in autism, we may to re-contact you to invite your ward/relative/person you are consenting for to participate in other studies. Participation in any such other studies is entirely voluntary - you can decide at the time. They will receive a maximum of 4 invitations per year to participate in other studies.

2.7 Will taking part in the study be kept confidential?
All the data we collect during the course of the research will be kept confidential and there are strict laws which safeguard the privacy of the patient at every stage. Study researchers will need access to the medical records of your ward/relative/person you are consenting for to carry out this research which is mentioned in detail below. We will store data and identifiers

on secure servers located at the University of Cambridge, details of which are provided below. To ensure that the study is being run correctly, we will ask your consent for responsible representatives from the Sponsor Cambridgeshire and Peterborough Foundation Trust and NHS Institution to access your medical records and data collected during the study, where it is relevant to you taking part in this research. The Sponsor is responsible for overall management of the study and providing insurance and indemnity.

2.8 What will happen if the ward/relative/person I am consenting for regains capacity during the course of the study?

If your ward/relative/person you are consenting for regains capacity during the course of the study, we ask you to please email us at tbc@spectrum10k.org to let us know and they will be asked to give their consent to continue in the study.

Any saliva sample provided or questionnaires you have completed on behalf of your ward/relative/person you are consenting for will be retained as part of their data collection within the study. If they regain capacity, any subsequent questionnaires/saliva sample that they provide will be added to this.

You will be directed to enter an email address for the participant who regains capacity. We will not collect any personal information about the person who regains capacity at this point; the email address will not be stored and will not be visible to us.

The person who regains capacity will receive a URL link which will direct them to register online. The account will be frozen at this point and you will no longer have access. The account will only be reactivated when the participant who regains capacity accesses the account and decides if they wish to continue in the study.

3. Data Use, Storage, and Safety

3.1 Summary

We will need to use information from your ward/relative/person you are consenting for and their medical records for this research project. This information will include their name, date of birth, NHS number, gender and postcode. People will use this information to do the research or to check their records to make sure the research is being done properly.

People who do not need to know who they are will not be able to see their name or contact details. The data of your ward/relative/person you are consenting for will have a code number instead. We will keep all information about them safe and secure. Once we have finished the study, we will keep some of the data so we can check the results. We will write our reports in a way that no-one can work out that they took part in the study.

Your ward/relative/person you are consenting for can stop being part of the study at any time, without giving a reason, but we will keep information about them that we already have. We need to manage their records in specific ways in order for the research to be reliable. This means that we won't be able to let you see or change the data we hold about them. If you agree to your ward/relative/person you are consenting for taking part in this study, they will have the option to take part in future research using their data saved from this study. You

can find out more about how we use your information at www.hra.nhs.uk/information-about-patients/

3.2 Will you have access to medical records?

One of the aims of Spectrum 10K is to identify factors that contribute to co-occurring physical and mental health conditions in autistic individuals. As part of this study we will apply for permission from the national dataset bodies to view participant electronic health records (EHRs). With your consent we will obtain health–related records and prescribed medicines held by the NHS General Practices and Hospitals of your ward/relative/person you are consenting for. This includes information about diagnosis (if applicable), hospitalisation records and test results (such as blood test and scan results). From these we will ensure a full understanding of the relationship between autism related genes, co-occurring conditions, medical care and outcomes. It will also help us to identify where current needs of autistic individuals and their families are not being met.

We will use details such as NHS number or equivalent, date of birth, name, gender and postcode to identify medical records. Data from these records will be shared with the Spectrum 10K research team and stored long-term. These data will be held under a unique study number assigned to your ward/relative/person you are consenting for, with all personal identifying information removed (i.e. pseudonymised). Contact details will be held in a separate database not linked to medical information or to the research data we hold about your ward/relative/person you are consenting for.

An application will be made to eDRIS (Scotland) who act as data controllers and who hold and maintain medical records and national datasets. For further information on the privacy policy of the organisation, please see the following:

eDRIS: https://www.isdscotland.org/Products-and-Services/eDRIS/How-eDRIS-is-Secure/

3.3 What will happen to the information of the ward/relative/person I am consenting for and how will you keep their data safe and confidential?

Once the steps are completed to sign up to Spectrum 10K, the data of your ward/relative/person you are consenting for will be stored within the Spectrum 10K database until they are withdrawn. Their data will be stored long-term by Cambridgeshire and Peterborough NHS Foundation Trust (CPFT) and the University of Cambridge for use in current and future research aimed to better understand autism, other co-occurring conditions, and wellbeing.

The data you provide for your ward/relative/person you are consenting for will be kept strictly confidential. All of their personal data will be stored on secure servers located at the University of Cambridge, and will only be accessible by approved Spectrum 10K researchers. All data shared with collaborators will be anonymised, meaning that your ward/relative/person you are consenting for will not be identified from any of the samples or data provided. The pseudonymised medical and health data of your ward/relative/person you are consenting for will be stored separately from their personal details. All data collected will be collected in line with the Data Protection Act 2018 and GDPR (General Data Protection Regulations). Data collection will also comply with the Common Law Duty of confidentiality.

CPFT and the University of Cambridge are joint sponsors for this study based in the United Kingdom. CPFT and the University of Cambridge will use data about your ward/relative/person you are consenting for and their medical records in order to undertake this study and will act as the data controller for this study. This means that they are responsible for looking after their

data and using it properly. With your consent, CPFT and the University of Cambridge will store identifiable and non-identifiable data long term. Data will be stored until you withdraw your ward/relative/person you are consenting for.

Your rights to access, change or move their data are limited, as we need to manage their data in specific ways in order for the research to be reliable and accurate. To safeguard the rights of your ward/relative/person you are consenting for, we will use the minimum personally-identifiable data possible.

You can find out more about how we use your data at: https://www.medschl.cam.ac.uk/research/privacy-notice-how-we-use-your-research-data/

3.4 What will happen to the saliva sample of my ward/relative/person I am consenting for?
The anonymised saliva sample of the ward/relative/person you are consenting for will be delivered to a laboratory to extract DNA for genetic analyses and long-term storage. We may analyse all or parts of your ward/relative/person's DNA.

3.5 What will your ward/relative/person's DNA be used for?
We will use your ward/relative/person's DNA to search for structural and functional changes in the DNA that may contribute to autism and related conditions. This will be done by analysing either their entire DNA code or only parts of their DNA code. We may also investigate how 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.

3.6 Who will analyse the data of my ward/relative/person I am consenting for?
The genetic and clinical data of your ward/relative/person you are consenting for will be analysed by Spectrum 10K team members or a trusted academic collaborator for ethically approved research.

3.7 Will data of my ward/relative/person I am consenting for be shared with academic researchers outside the Spectrum 10K study?
We may share the anonymized (genetic and questionnaire) data of your ward/relative/person you are consenting for using highly secure research databases (repositories) or share with potential academic collaborators for future research. We will not share any personal data with other researchers.

3.8 Will data of my ward/relative/person I am consenting for be shared with commercial collaborators?
In some instances, Spectrum 10K may also share anonymised data with commercial collaborators. **This is optional**, and you can indicate if you would like the data of your ward/relative/person you are consenting for to be shared. Any commercial collaboration will be in line with the aims of Spectrum 10K. Some examples of such collaborations may include developing a drug for a specific type of epilepsy or gut difficulties relevant to some autistic individuals, or developing an algorithm to better detect depression in autistic individuals. Such research is typically conducted in commercial collaborations.

3.9 What if the ward/relative/person I am consenting for does not want to continue in the study?

If you think your ward/relative/person you are consenting for would no longer wish to take part in the study, you are free to withdraw them from Spectrum 10K at any time. You do not have to give a reason and the care provided to the ward/relative/person you are consenting for will not be affected in any way. You are welcome to discuss concerns with us at any time, and the various options you have for withdrawal. Please contact the Spectrum 10K team by phone on xxxxxxx or email the team at tbc@spectrum10k.org.

You have three options for the withdrawal of the individual you are consenting for:

No Further Contact:

This means that Spectrum 10K would no longer contact you directly but would have permission to retain and use information and samples provided previously by your ward/relative/person you are consenting for, and to obtain and use further information from the health records of your ward/relative/person you are consenting for. This level of withdrawal allows researchers to study autism with the goal of improving the health of people with autism and their family members.

No Further Access:

This means that Spectrum10K would no longer contact you or obtain further information from the health records of your ward/relative/person you are consenting for in the future; but still has permission to use the information and samples provided previously.

No Further Use:

In addition to no longer contacting you or obtaining further information, any information and samples collected previously would no longer be available to researchers. Spectrum 10K would destroy the sample of your ward/relative/person you are consenting for (although it may not be possible to trace all distributed sample remnants) and would only hold information for archival audit purposes. Such a withdrawal would prevent information about your ward/relative/person you are consenting for, from contributing to further research, but it would not be possible to remove data from research that had already taken place

3.10 Who has reviewed the study?

Spectrum 10K has been peer reviewed. All research in the NHS is looked at by an independent group of people, called a Research Ethics Committee. A favourable ethical opinion has been obtained from Scotland A-REC. NHS management approval has also been obtained.

4. Advantages and Disadvantages

4.1 What are the possible benefits of taking part?

Your ward/relative/person you are consenting for will not benefit directly from taking part in Spectrum 10K. However, the information we gain from this study will help to further our understanding of the relationship between genes, autism and related conditions. This research may lead to improved quality of care for autistic individuals and their families.

4.2 What are the possible disadvantages and risks of taking part?

1. Providing a saliva sample is a very low risk procedure but may be unpleasant for some people. We will offer foam swabs to aid the collection of saliva to participants who cannot

produce enough saliva through spitting. You will have the option to request the assisted saliva collection kit during registration.

2. Our questionnaires may cover topics which some people might find sensitive, and about difficult lived experiences of mental or physical conditions. Although this relates to your ward/relative/person you are consenting for, this may affect you too. You will have the option to skip some questions by selecting the 'I prefer not to answer' option.

3. The participation of your ward/relative/person you are consenting for is strictly confidential. Where appropriate, we may share their anonymised data in highly secure research databases or share with potential collaborators. The risk of identifiable information being accidentally disclosed is extremely low.

4.3 Will Spectrum 10K provide feedback about the genetic results of my ward/relative/person I am consenting for?

We understand that some autistic individuals and their family members (or carers) would like to receive feedback about their DNA. However, we do not currently have the facilities to provide feedback on individuals' genetic data in an ethical manner that minimizes distress with the right support and counselling services. For this reason, we are currently unable to provide feedback on genetic data. However, we may consider developing an infrastructure to do so in the future. If this becomes possible, we will contact all participants in the future requesting consent to provide feedback to them about their genetic data.

However, your ward/relative/person you are consulting for may be eligible for genetic testing in certain situations. These may include having a developmental delay, a learning disability, and congenital abnormalities. If you think they might be eligible for genetic testing, please consult their GP to find out how to get a referral.

4.4 Will you contact the GP of my ward/relative/person I am consenting for?

In some exceptional circumstances, we may need to contact the GP of your ward/relative/person you are consenting for to inform them about their participation in the study and any important information related to their health and safety that we hold about them. This will occur only if we find something that may affect their clinical care. Please note, we will not be contacting their GP routinely to inform them about the participation in the study nor will we be routinely feeding back any information you provide us to the GP of your ward/relative/person you are consenting for.

5. Final Details

5.1 Will we be in contact during this study?

As part of Spectrum 10K, we will send reminder emails and updates about the participation and progress of the ward/relative/person you are consenting for.

Additionally, you will be given the option to subscribe to our newsletter. If you are interested in receiving our newsletter, please consent to be on our mailing list. You will receive a maximum of 2-3 newsletters per year and are free to withdraw from the mailing list at any time by emailing the research team at TBC@spectrum10k.org.

5.2 What if there is a problem?

If you have a concern about any aspect of this study please contact the research team at tbc@spectrum10K.org who will do their best to answer your questions. In the unlikely event that something goes wrong and the ward/relative/person you are consenting for is harmed during the research and this is due to someone's negligence then you may have grounds for a legal action for compensation against the NHS but you may have to pay your legal costs.

The normal National Health Service complaints mechanisms will still be available to you (if appropriate).

5.3 What happens at the end of Spectrum 10K?
The study results will be presented at research conferences and written up in scientific journals. The ward/relative/person you are consenting for will not be identifiable in any published results.

5.4 Who is organising the research and why?
This study has been jointly organised/sponsored by Cambridgeshire and Peterborough NHS Foundation Trust and the University of Cambridge, and is funded by the Wellcome Trust.

If you have any further questions about the study please contact the Spectrum 10K research team **on: (TBC) or email: info@Spectrum10k.org**

If you have any concerns or issues and would like to discuss this study with someone independent of the study, please email the designated independent contact Professor Tony Holland at: tonyholland@medschl.cam.ac.uk

If you wish to make a complaint about the study please contact NHS Lothian:

NHS Lothian Complaints Team
2nd Floor
Waverley Gate
2 - 4 Waterloo Place
Edinburgh
EH1 3EG
Tel: 0131 465 5708
craft@nhslothian.scot.nhs.uk

Thank you for taking the time to read this information sheet