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Participant Information Sheet
(Adult Participant)
Study Title: Spectrum 10K

REC Ref: **20/LO/0873**
IRAS Ref: 266324

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

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 professional diagnosis of Autism Spectrum Conditions (hereafter 'autism') from a qualified professional and, where possible, their relatives to identify genetic and environmental factors that contribute to autism and related conditions. This will help us to accelerate understanding of the biological and environmental factors that contribute to the wellbeing of autistic individuals.

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 to take part, completing questionnaires, providing a saliva sample and providing access to your medical records (to investigate important clinical information relevant to the study). Participation in this study is completely voluntary. You can withdraw at any time without giving a reason.

2. Taking Part in Spectrum 10K

2.1 Why have I been invited to participate?

We are inviting you to participate because you have seen the study advertised in the press and/or on social media, or have been invited by a relative, or have been approached by your clinician, and you have indicated that:

- You are aged 16+ and have a diagnosis of autism from a qualified professional.

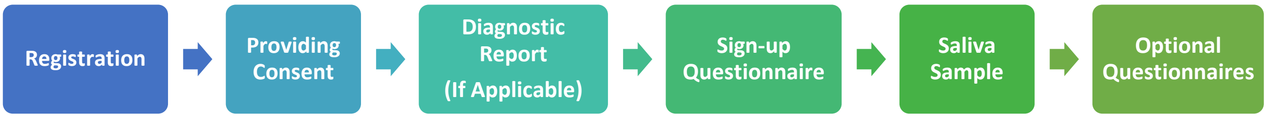
OR

- You are aged 16+ and are the biological parent of an autistic child below the age of 16 who is participating in Spectrum 10K.
  OR
- You are aged 16+ and are 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-grandparent).

2.2 Do I have to take part in Spectrum 10K?

No, your participation in Spectrum 10K is voluntary. There is no obligation to take part, and you can withdraw from Spectrum 10K at any time without giving a reason, and without your medical care or legal rights being affected.

2.3 What will my participation involve?



Step 1. Registration: If you decide to register for Spectrum 10K at www.spectrum10k.org, you will be asked to complete the following:

- **Consent:** You will be asked to complete an online consent form which outlines the tasks you may be involved in. Once you give your consent, 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, you will not be excluded from the study, however all other consent points are required for study participation.

- **Participant Information:** You will be asked to provide your name, date of birth, email address, home address, information about your autism diagnosis (if applicable), and other basic information about yourself. We will also ask you to provide, if possible, your NHS number to link to your Electronic Health Records. Please see below for more information about collecting information about your medical records.

- **Clinical Report (If Applicable)**: If you have an autism diagnosis from a qualified professional, you will be asked to upload a copy of your report with information about your 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. Your 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 your 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 your autism diagnosis.

- **Sign-up Questionnaire (Baseline Questionnaire):** You will be asked to complete a sign-up questionnaire. This includes questions about your health, lifestyle, education and questions related to autistic traits. You will need to complete this questionnaire even if you are not autistic. This usually takes 20 – 25 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. This is optional and will not stop you participating in Spectrum 10K if you do not want to complete it. The questionnaire will ask about whether you were unwell due to COVID-19 or knew a person who was, symptoms you experienced, treatment management and shielding. It will also ask about the impact of COVID-19 on your finances, mood and anxiety levels. This usually takes 25 – 30 minutes. If you choose to complete the COVID-19 questionnaire, we will ask you to repeat the questionnaire every 3 months for 2 years. This is so we can study the long-term effects of the pandemic on autistic individuals and their families. You will receive an email reminding you to complete the questionnaire when it is due.

Step 2 - Saliva DNA Kit: We will send you a spit tube to provide your saliva sample, instructions on how to do so will be included along with pre-paid return envelopes. We will extract DNA from your sample.

To use the saliva kit all you will need to do is spit into the tube. If you think you might have problems producing a saliva sample through spitting, please request an assisted DNA saliva kit with foam swabs during registration.

Step 3: Optional Questionnaires:
You will have the option to log in to your Spectrum 10K account (at www.spectrum10k.org) and complete additional questionnaires. There are 5 sets (or phases) of optional questionnaires covering a range of different aspects of autism, co-occurring conditions and wellbeing. Each phase usually takes 20-25 minutes to complete. 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 phase over a period of 2 years after you register.

Optional questionnaires will include sensitive questions related to your mental and physical health and include potentially distressing topics such as abuse and suicide which you 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 members of my family participate in Spectrum 10K?

Yes, if you have a professional autism diagnosis you can invite your biological relatives (up to third-degree) to participate in Spectrum 10K. Third degree eligible relatives include your: biological parents, biological children, biological (half or full) siblings, biological aunts and uncles, grandparents, first cousins and great-grandparents.

If you do not have an autism diagnosis, then you will not have the option to invite these relatives.

If eligible, you will have an option to invite your relatives to the study once you have completed your own registration. You can register children under your care using the same account. For relatives not under your care, you will need to specify your relationship to each individual and enter their email address. Your relatives will receive a URL link which will direct them to register online. Using this URL method will allow for your relatives to be automatically linked to you. We will not collect any personal information about your relatives at this point; the email address will not be stored and will not be visible to us.

The relatives you invite to 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 be paid?

Unfortunately, we are not able to pay you for your participation in this study.

2.6 Will I be re-contacted?

To better understand co-occurring conditions and wellbeing in autism, we may re-contact you to invite you to participate in other ethically approved studies. Participation in any such other studies is entirely voluntary - you can decide at the time. You will receive a maximum of 4 invitations per year to participate in other studies.

3. Data Use, Storage, and Safety

3.1 Summary

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

People who do not need to know who you are will not be able to see your name or contact details. Your data will have a code number instead. We will keep all information about you 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 you took part in the study.

You can stop being part of the study at any time, without giving a reason, but we will keep information about you that we already have. We need to manage your 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 you. If you agree to take part in this study, you will have the option to take part in future research using your 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 my 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 your electronic health records (EHRs). With your permission and consent we will obtain information about your health and prescribed medicines from your records held by the NHS General Practices and Hospitals. This includes information about any diagnoses you may have, your hospitalization records, and your test results (such as blood test and scan results). From these, we will obtain 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 your details such as your NHS number or equivalent, date of birth, name, gender and postcode to identify your 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 you, with all personal identifying information removed (i.e. pseudonymised). Your contact details will be held in a separate database not linked to your medical information or to your research data.

An application will be made to the national bodies NHS Digital (England), SAIL database (Wales), NSS/eDRIS (Scotland), HSC Trust (Northern Ireland) who act as data controllers, and who hold and maintain medical records and national datasets. For further information on the privacy policy of these organisations, please see the following:

NHS Digital: https://digital.nhs.uk/about-nhs-digital/our-work/keeping-patient-data-safe/how-we-look-after-your-health-and-care-information

SAIL Databank: https://saildatabank.com/saildata/data-privacy-security/

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

NSS: https://nhsnss.org/how-nss-works/policies-and-statements/privacy-advisory-committee/

HSCNI: https://www.health-ni.gov.uk/sites/default/files/publications/dhssps/hsc-security-policy.docx

3.3 What will happen to my information and how will you keep my data safe and confidential?

Once you have completed the steps to sign up to Spectrum 10K, your data will be stored long-term within the Spectrum 10K database. Your 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 ethically approved research aimed to better understand autism, other co-occurring conditions, and wellbeing.

The data you provide will be kept strictly confidential. All of your 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 you will not be identified from any of the samples or data you provide. Your pseudonymised medical and health data will be stored separately from your 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 of this study based in the United Kingdom. CPFT and the University of Cambridge will use data about you and your medical records in order to undertake this study and will act as the data controller for this study. This means that we are responsible for looking after your data and using it properly. With your consent, CPFT and the University of Cambridge will store identifiable and non-identifiable data long term. We will keep data until you choose to withdraw.

Your rights to access, change or move your data are limited, as we need to manage your data in specific ways in order for the research to be reliable and accurate. To safeguard your rights, 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 my saliva sample?

Your anonymised saliva sample will be delivered to a laboratory to extract DNA for genetic analysis. We may analyse all or parts of your DNA. Your DNA sample will be stored long-term.

3.5 What will my DNA be used for?

We will use your 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 your entire DNA code or only parts of your DNA code. 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.

3.6 Who will analyse my data?

Your genetic and clinical data will be analysed by Spectrum 10K team members or a trusted academic collaborator for ethically approved research.

3.7 Will my data be shared with academic researchers outside Spectrum 10K?

We may share your anonymized (genetic and questionnaire) data 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 my data 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 your data to be shared. Any commercial collaborations 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 will happen if I want to withdraw from the study?

You are free to withdraw from Spectrum 10K at any point without giving us a reason. You are welcome to discuss concerns with us at any time, and the various options you have for withdrawal. If you do wish to withdraw, please contact the Spectrum 10K team by phone on 0800 052 0050 or email the team at info@spectrum10k.org.

You have three options for withdrawal:

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 you provided previously and to obtain and use further information from your health records. 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 health records 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 samples (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 you 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 reviewed and a favourable opinion was given by independent ethics review committees at the London Queens Square REC & Scotland A-REC..

4. Advantages and Disadvantages

4.1 What are the possible benefits of taking part?

There are no direct benefits of taking part. However, Spectrum 10K 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 of taking part?

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

2. Our questionnaires may cover topics which some people might find sensitive, and about difficult lived experiences of mental or physical conditions. You will have the option to skip some questions by selecting the ‘I prefer not to answer’ option.

3. Your participation is strictly confidential. Where appropriate, we may share your 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 my genetic results?

We understand that some autistic individuals and their family members would like to receive feedback about their DNA. However, we do not currently have the facilities to provide feedback on your 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 your 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 you about your genetic data.

You 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 you might be eligible for genetic testing, please consult your GP to find out how to get a referral.

4.4 Will you contact my GP about my participation in this study?

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

5. Final Details

5.1 Will you contact me during this study?

As part of Spectrum 10K, you will receive reminder emails and updates about your participation and progress. 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 info@spectrum10k.org.

5.2 What happens at the end of Spectrum 10K?

Results will be presented at research conferences and written up in scientific journals. The data will be totally anonymous, without any means of identifying the individuals involved.

5.3 What if there is a problem?

If you have a concern about any aspect of this study please contact the research team at info@spectrum10K.org who will do their best to answer your questions. In the unlikely event that something goes wrong and you are 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 (PALS) will still be available to you (if appropriate).

6. Contact Spectrum 10K

If you have any concerns or need further details before consenting to take part, please contact our research team at info@spectrum10k.org. If you wish to participate, please continue your online registration at www.spectrum10K.org. If you do not wish to participate, then you do not have to do anything.