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Participant Information Sheet
(Parent for Child)
Study Title: Spectrum 10K

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This information sheet will explain why this research is being done and what your child's 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 your child up to Spectrum 10K (at www.spectrum10k.org) will involve giving consent to take part, completing questions about your child, obtaining a saliva sample from your child, and consenting to our team accessing your child's medical records (to investigate important clinical information relevant to the study). Participation in this study is completely voluntary. You can withdraw your child from the study at any time without giving a reason.

2. Taking Part in Spectrum 10K

2.1 Why has my child been invited to participate?

We are inviting your child 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 child's clinician, and you have indicated that you are the parent or legal guardian of:

- A child below the age of 16 with a diagnosis of autism from a qualified professional.
OR
- A child below the age of 16 who 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).

2.2 Does my child have to take part in Spectrum 10K?

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

2.3 What will my child's participation involve?



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

- **Consent:** You will be asked to complete an online consent form which outlines the tasks your child may be involved in. If your child turns 16 years of age during the study period, we will contact you to ask you to invite your child to provide their own consent to continue being part of Spectrum 10K. If your child is without capacity when they turn 16, we will ask if you are willing to act as a consultee to provide advice on their behalf.

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

If your child is 6 years of age or over, does not have a diagnosis of intellectual disability, and you indicate that they can read and understand information appropriate for their age, we will additionally provide adapted information sheets. These documents will provide your child with information about the study in an easier read format and explain what is involved in taking part. Following research ethics guidelines, we will ask that your child completes an 'assent form' which asks 'yes' or 'no' questions to confirm they are happy to participate. This is not obligatory but we recommend that this step is completed.

- **Participant Information:** As the parent or legal guardian you will be asked to provide your name, your date of birth, your email address, and home address, to allow us to contact you and your child. You will then be asked to provide your child's name, your child's date of birth, information about your child's diagnosis and other basic information.

We will also ask you to provide, if possible, your child's NHS number to link to your child's Electronic Health Records. Please see below for more information about collecting information about your child's medical records.

- **Clinical Report (If Applicable):** If your child has an autism diagnosis from a qualified professional, you will be asked to upload a copy of your child's clinical report with information about your child's 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 child's diagnostic 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 child's 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 child's autism diagnosis.

- **Sign-up Questionnaire (Baseline Questionnaire):** You will be asked to complete the sign-up questionnaire about your child. This includes questions about your child's health, lifestyle, education and questions related to autistic traits. You will need to complete this questionnaire even if your child

is not autistic. We may ask you to consult your child on some questionnaires if possible or ask you to ask your child to complete a few questionnaires if possible (such as a memory test). 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 about your child. This is optional and will not prevent your child participating in Spectrum 10K. The questionnaire will ask about whether your child was unwell due to COVID-19 or knew a person who was, symptoms your child experienced, treatment management and shielding. It will also ask about the impact of COVID-19 on your child's 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 that 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 for you to collect your child's saliva sample, instructions on how to do so will be included along with pre-paid return envelopes. We will extract DNA from your child's sample.

To use the saliva kit all you will need to do is get your child to spit into the tube. If you think your child 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 about your child. 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 child's 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 your child has a professional autism diagnosis, you can invite your child's biological relatives (up to third-degree) to participate in Spectrum 10K. Third degree eligible relatives include: biological parents, biological children, biological (half or full) siblings, biological aunts and uncles, grandparents, first cousins and great-grandparents.

If your child does 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 child's relatives to the study once you have completed your child's registration. If you are the biological parent, you can register yourself and other children (below the age of 16) under your care using the same account. For relatives not under your care, you will need to specify your child's 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 your child. 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 or my child be paid?

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

2.6 Will my child be re-contacted?

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

3. Data Use, Storage, and Safety

3.1 Summary

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

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

Your child can stop being part of the study at any time, without giving a reason, but we will keep information about your child 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 your child. If your child agrees to take part in this study, your child will have the option to take part in future research using data saved from this study.

You can find out more about how we use your child's information at:

www.hra.nhs.uk/information-about-patients/

3.2 Will you have access to my child's 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 child's electronic health records (EHRs). With your permission and consent we will obtain information about your child's health and prescribed medicines from your child's records held by the NHS General Practices and Hospitals. This includes information about any diagnoses your child may have, hospitalization records, and 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 are not being met.

We will use your child's details such as their NHS number or equivalent, date of birth, name, gender and postcode to identify their medical records. Data from these records will be shared with the Spectrum 10K research team. These data will be held under a unique study number assigned to your child, with all personal identifying information removed (i.e. pseudonymised). Your contact details will be held in a separate database not linked to your child's medical information or to their 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://nhs.uk/healthcare/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 child's information and how will you keep my child's data safe and confidential?

Once you have completed the steps to sign your child up to Spectrum 10K, your child's data will be stored within the Spectrum 10K database. Your child's data will be stored 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 about you or your child will be kept strictly confidential. All of your personal information will be stored on secure servers located at the University of Cambridge, and will only be accessible by approved researchers from Spectrum 10K. All data shared with collaborators will be anonymised, meaning that your child will not be identified from any of the samples or data you provide. 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 child, and your child's 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 child's data and using it properly. With your consent, CPFT and the University of Cambridge will store identifiable and non-identifiable data about you and your child long term. We will keep data until you choose to withdraw your child from the study.

Your rights to access, change or move your child's data are limited, as we need to manage your child's data in specific ways in order for the research to be reliable and accurate. To safeguard your child's rights, we will use the minimum personally-identifiable data possible.

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

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.

3.4 What will happen to my child's saliva sample?

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

3.5 What will my child's DNA be used for?

We will use your child'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 your child's entire DNA code or only parts of your child's DNA code. We may also investigate how your child's 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 child's data?

Your child's 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 child's data be shared with academic researchers outside Spectrum 10K?

We may share your child's 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 child's 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 child's 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 my child from the study?

You are free to withdraw your child 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 your child, 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 your child's 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 your child provided previously and to obtain and use further information from your child's 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 your child's 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 your child's sample (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 child 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 London Queens Square REC and 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 child's participation is strictly confidential. Where appropriate, we may share your child's 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 child's 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 child's 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 child's 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.

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

4.4 Will you contact my child's GP about their participation in this study?

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

6 Contact Spectrum 10K

If you have any concerns or need further details before consenting for your child to take part, please contact our research team at info@spectrum10k.org. If you wish to register your child to participate,

please continue your registration at www.spectrum10K.org. If you do not wish for your child to participate, then you do not have to do anything.