Title of Study: DELineating the NEuropsychologicAl phenoTypEs of rare developmental genetic conditions (DELINEATE)

Parent / Guardian Information Sheet

We would like to invite your child to take part in a research study looking at the cognitive, behavioural, emotional, and social skills (abilities that are collectively known as “neuropsychological functioning”) of individuals who are affected by certain rare conditions. This study is being carried out as part of a PhD programme at the University of Manchester.

Before you decide whether or not for your child to take part, it is important that you understand why the research is being done, and what it will involve. Please take time to read the following information carefully before deciding whether to allow your child to take part and to 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 in advance for taking the time to read this.

About the research

Who is carrying out the research?

I am Aoife O’Sullivan, a PhD student in the Division of Neuroscience at the University of Manchester. I will be conducting the study as part of a research team under the supervision of my academic supervisors: Dr Rebecca Bromley and Professor Siddharth Banka, both of whom are based at the University of Manchester and at Manchester University NHS Foundation Trust.

What is the purpose of this research?

Some genetic conditions can change the way a person’s brain develops and the way it functions. Many of these conditions are very rare and are not well understood. We would like to learn more about the neuropsychological functioning (i.e., thinking skills, behaviours, emotions, and social abilities) associated with different rare developmental genetic conditions to better understand how they impact on a person’s daily life.

We would also like to learn the best way to measure these skills in people who are living with rare developmental genetic conditions.

We are hoping to recruit up to 200 people who are living with a rare developmental genetic condition to take part in our research.

Who can take part in this study?

We are inviting people with rare developmental genetic conditions to take part in our study.

If you are unsure whether this applies to your child, please contact us using the details at the bottom of this information leaflet and we would be happy to talk to you about it.

What if my child has already completed a neuropsychological assessment in the past?

If your child has previously completed a neuropsychological assessment as part of their routine NHS clinical care then they may not need to do another assessment as part of this study. Instead, we will ask you to share their old neuropsychological reports with us and for your consent for us to include this data in our analysis.

We may invite your child to complete an updated neuropsychological assessment if it has been more than 24 months since they were previously assessed.

What if English is not my or my child's native language?

This is not a problem and does not exclude your child from our study. The Manchester University NHS Foundation Trust Interpretation & Translation Service can be used to facilitate conversations if your or their first language is not English.

What will happen to the results of this study?

The results will be reported in full in the PhD thesis produced by Aoife O'Sullivan (but participants will not be identified by name).

Our findings will also be published in academic journals, included in presentations to other healthcare professionals, and shared at conferences.

The overall results of the study will be shared with families to have taken part and relevant charities and patient groups.

Disclosures and Barring Service (DBS) Check

All researchers have undergone an enhanced DBS check obtained via The University of Manchester, or another external organization.

Who is funding the study?

The study is being undertaken as part of a PhD studentship that is fully funded by the **National Institute for Health and Care Research Manchester Biomedical Research Centre**.

Who has reviewed the study?

The study has been approved by an independent group of people, called a **Research Ethics Committee** (REC 25/YH/0024 / IRAS 345536), whose job is to protect your safety, rights, wellbeing, and dignity.

What would my child's involvement be?

What would my child be asked to do if they took part?

Consent
If you decide for your child to take part in our study after reading this information sheet, the first thing we will ask you to do is to complete a consent form. This will include asking you to consent to your child taking part in a neuropsychological assessment, if necessary.

Access to medical records
You will also be asked to consent to a member of the research team viewing your child's medical notes. We want to learn about their medical history so that we can plan appropriately for their neuropsychological assessment.

Collection of medical history
With your consent, a member of the research team will review your child's medical records to collect information about their early life development, their diagnosis, and the symptoms they have experienced. We may ask you some questions about your child's medical history if we cannot access their records directly.

Collection of demographic details
With your consent, we will also collect some general information about your child including their name, gender, date of birth, ethnicity, and contact details. We need this information to communicate with you and to understand your child's assessment results.

Appointment Booking
Next, a member of the research team will contact you to arrange a time to complete your child's neuropsychological assessment. We will also make a plan for their assessment at this time. We will decide whether the assessment will be done face-to-face or by video call and whether your child will need a translator to help them.

Neuropsychological Assessment
The neuropsychological assessment will then take place at a location convenient to you and your child. The appointment will last a maximum of 2.5 hours, with plenty of breaks given as needed. We may need a second appointment to complete the assessment.

Remote Neuropsychological Assessment
Some people will complete an assessment over video call. These assessments will be shorter than the face-to-face assessments, lasting one hour in duration.

Questionnaires
We will also ask you to complete questionnaires about your child's behavioral, emotional, sensory, and social development. They may also be asked to complete a copy of some of these questionnaires.

What would I be asked to do if I took part? (continued)

Feedback

We will send you some short feedback about how your child performed on different tasks over the course of their neuropsychological assessment within eight weeks of their appointment.

End of Study

Once we have made sense of the results of all of the assessments completed as part of this study, we will send you a summary of what we have found.

What will my child be asked to do during their neuropsychological assessment?

Neuropsychological assessments will be carried out to learn more about your child's cognitive (thinking) skills. This will be done by asking them to complete a range of puzzles, tasks, and problems. There are no right or wrong answers to any of these tasks, we just want to learn more about how your child's brain works. We will also have some questions for you to answer as their primary caregiver.

The assessment may be completed either at the Royal Manchester Children's Hospital, at your child's school/college (if they are still attending one), at home, or over video call. The appointment will last up to 2.5 hours, including breaks.

We may need to arrange a second appointment to complete the assessment if we do not get everything finished in the first appointment.

Will my child be compensated for taking part?

Unfortunately, participants will not be financially compensated for taking part in this study.

Are there any benefits to taking part?

We will provide you with a brief written summary of your child's neuropsychological assessment scores. These results will help you to understand your child's personal strengths and can guide decision-making about the type and level of support that they may benefit from having access to in their daily life.

The overall results of the study will improve our understanding about how the brains of individuals with rare developmental genetic conditions work. This means that healthcare professionals will able to offer better advice and support regarding the management of these conditions in the future.

Are there any risks to taking part?

The tests and questionnaires that we will ask your child to complete will be the same as what is undertaken in a routine clinical assessment. However, assessing your child's physical and neurodevelopment in this way may highlight areas where they may have a particular difficulty or weakness. In this situation, we will encourage you to contact your child's GP and to discuss any concerns that you may have with them. It may be helpful to send your child's GP a copy of their neuropsychological assessment results too.

What happens if I do not want my child to take part or I change my mind?

It is up to you to decide whether or not for your child to take part. You can tell us if you wish for them to take part by sending an email to **aoife.osullivan@postgrad.manchester.ac.uk** or by scanning the QR code at the bottom of this leaflet.

If you do decide for your child to take part, you will be given this information sheet to keep and will be asked to sign a consent form. If you decide for your child to take part you are still free to withdraw them at any time without giving a reason and without harm to them or any impact on their NHS care. However, it will not be possible to remove your child's data from the project once it has been combined with the overall dataset for statistical analysis as we will not be able to identify their specific data. This does not affect their data protection rights.

If you decide for your child not to take part, you do not need to do anything further.

Does my child have to take part?

No. Participation is completely voluntary and choosing not to take part will not affect your child's medical care in any way.

Data Protection and Confidentiality

What information will you collect about my child?

In order to participate in this research project, we will need to collect information that could identify your child, called **“personal identifiable information”**. Individuals from the University of Manchester and Manchester University NHS Foundation Trust will access your child’s medical records to collect information for the study. Specifically, we will need to collect the following information about your child:

Name | NHS/HSC/CHI number | Gender | DOB | Ethnicity | Diagnosis | Symptoms | Health information | Developmental History | Postal address | Contact details

We will use this information to carry out our research. People may also use this information to check your child’s records to make sure that the research is being done properly. People who do not need to know who your child is will not be able to see their name or contact details. Their data will have a code number instead. We will keep all information about your child safe and secure in a password protected database called **REDCap**.

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.

Under what legal basis are you collecting this information?

We are collecting and storing this personal identifiable information in accordance with UK data protection law which protect you and your child’s rights. These state that we must have a legal basis for collecting your child’s data. For this study, the specific reason is that it is **“a public interest task”** and **“a process necessary for research purposes”**.

What are my rights in relation to the information you collect about my child?

You have a number of rights under data protection law regarding your child’s personal information. For example, you can request a copy of the information we hold about your child, including test scores. If you would like to know more about your different rights or the way we use your child’s personal information to ensure we follow the law, please consult the “Privacy Notice for Research Participants” which is available here: https://documents.manchester.ac.uk/display.aspx?DocID=37095.

Will my child's participation in the study be confidential and their personal identifiable information be protected?

In accordance with data protection law, the University of Manchester is the **Data Controller** for this project. This means that we are responsible for making sure your child's personal information is kept secure, confidential, and used only in the way you have been told it will be used. All researchers are trained with this in mind, and your child's data will be looked after in the following way:

- **Personal information:** The study team at The University of Manchester will have access to your child's **personal information** and they will pseudonymise it as soon as possible. Your child's name and any other identifying information will be removed and replaced with a unique Study ID number. The research team will have access to the key that links this ID number to your personal information. With your consent, we would also like to retain your **contact details** for 5 years in order to provide you with a summary of the findings for this study and also to inform you about future studies that you and your child may be interested in. If you provide consent for this, your details will be safely stored on University of Manchester servers in a digital folder only accessible to the study team and used only for the purposes described above. If you do not wish to be contacted about the results of the current study or to hear about future research, your contact details will be deleted immediately upon completion of the study.
- **Consent Form:** The consent form you sign on your child's behalf will be retained for 2 years in a locked cabinet on UoM premises for audit purposes.
- **Study Data:** We will enter the health and developmental information we collect about your child from their medical notes and their neuropsychological test scores into our secure **REDCap** research database, held at the University of Manchester. The information will be stored according to a Study ID number unique to your child and not stored by their name or other identifiable information. The key linking your child to their Study ID will only be accessible to core members of the research team. The key linking each participant to their unique Study ID will be destroyed at the end of the current study to fully anonymise the data.
- **Use of Zoom and/or Microsoft Teams:** Remote neuropsychological assessments will be completed using these platforms. These assessments will not be recorded and so participant data will not leave the European Economic Area.
- **Data Archiving:** At the end of the project, we will transfer the fully anonymised REDCap dataset to the University of Manchester's **Research Data Storage** service where it will be held for 10 years. We will not be depositing our research data into an open repository for permanent storage in order to protect the identities of our participants, who may be recognised from their clinical features.

Will my child's participation in the study be confidential and my child's personal identifiable information be protected? (continued)

- **Case Studies:** It may happen that we need to consider publishing a case study or report to inform the medical community of important information relating to a particular individual, their diagnosis, and their pattern of neuropsychological functioning. Sharing this information will help us to better understand the strengths and weaknesses unique to certain conditions. Should we find something unusual or noteworthy in your child's assessment performance, we will discuss it with you and outline the information that we would like to publish. We will only publish a case study with your explicit consent.
- **Data Sharing:** When you agree to take part in a research study and with your informed consent, the information about your child may be provided to researchers running other studies here or at other organisations. These organisations may be universities, NHS organisations, or companies involved in health and care research in this country or abroad. Your child's information will only be used by organisations and researchers to conduct research in accordance with the **UK Policy Framework for Health and Social Care Research** and The University of Manchester's Research Privacy Notice. This information will not identify your child and will not be combined with other information in a way that could identify them. The information will only be used for the purpose of improving our understanding of the neuropsychological functioning associated with different rare developmental genetic conditions and cannot be used to contact you or your child regarding any other matter. It will not be used to make decisions about future services available to your child. This data may be analysed anonymously in either the UK or a different country. In these cases, they must follow our rules about keeping your child's information safe.
- **Loss of capacity:** Your child's continued capacity to take part in this study will be judged at every contact point. If any concerns arise, we will speak to you and their care team. If necessary, your child will be withdrawn from the study.
- Please also note that individuals from The University of Manchester, Manchester University NHS Foundation Trust, or regulatory authorities may need to look at the data collected for this study to make sure the project is being carried out as planned. This may involve looking at identifiable data, but all individuals involved in auditing and monitoring the study will have a strict duty of confidentiality to your child as a research participant.

Will my child’s data be used in future research studies?

If you consent to it, your child’s data may be used in a pseudonymous (de-identified) form in future ethically approved research conducted by this study team. For example, we may run more studies in the future that will improve our understanding of the neuropsychological skills associated with different rare developmental genetic conditions.

With your permission, we may also share anonymised study data with third parties such as commercial organisations (e.g., pharmaceutical companies developing or trialling a therapy for rare developmental conditions) or patient support organisations (e.g., for use in developing patient information guides).

Disclosures

If, during the study, we have concerns about your child’s safety or the safety of others, we will inform their care team.

If, during the study, you or your child disclose information about any current or future illegal activities, we have a legal obligation to report this and will therefore need to inform the relevant authorities.

If, during the study, concerns arise regarding your child’s clinical care we will flag this with the Chief Investigator who will then liaise with your child’s care team.

Individuals from the University, the site where the research is taking place and regulatory authorities may need to review the study information for auditing and monitoring purposes or in the event of an incident.

A court can, in exceptional circumstances, order researchers to disclose confidential information that they have collected as part of research projects. If a court orders disclosure of information collected from your child, confidentiality can no longer be maintained.

What if I have a complaint?

Contact details for complaints

If you have a complaint that you wish to direct to members of the research team, please contact:

Dr Rebecca Bromley
Research Fellow and Paediatric Neuropsychologist
via email at rebecca.bromley@manchester.ac.uk

Contact details for formal complaints

If you wish to make a formal complaint to someone independent of the research team or if you are not satisfied with the response you have gained from the researchers in the first instance then please contact:

The Research Ethics Manager, Research Office, Christie Building, The University of Manchester, Oxford Road, Manchester, M13 9PL
via email at research.complaints@manchester.ac.uk
or by telephoning 0161 306 8089

If you wish to contact us about your data protection rights, please email dataprotection@manchester.ac.uk or write to **The Information Governance Office, Christie Building, The University of Manchester, Oxford Road, M13 9PL** at the University and we will guide you through the process of exercising your rights.

You also have a right to complain to the **Information Commissioner's Office about complaints relating to your personal identifiable information** (https://ico.org.uk/make-a-complaint/). They can be contacted by telephoning **0303 123 1113**.

Contact Details

I have a question or would like to talk this through with someone?

We would be more than happy to respond to any questions or queries that you have about any element of this research project. If you have any queries about the study or if you are interested in your child taking part then please contact the researcher:

Ms Aoife O'Sullivan
via email at aoife.osullivan@postgrad.manchester.ac.uk

Alternatively, you can scan the QR code below and this will take you to the online consent form:

We really appreciate your participation in this important project.