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'ACORN' Study

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Chief Investigator: Dr Alexander G. Thompson

Participant Information Sheet – Study Partner

You are being invited 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. Please take time to read the following information carefully. Talk to others about the study if you wish.

This document tells you the purpose of this study and what will happen to you if you take part as well as more detailed information about the conduct of the study.

Ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part. We are planning to recruit participants into this study for five years, so you can decide to participate later on if this is more convenient.

What is the purpose of the study?

The purpose of this study is to investigate possible causes of nerve damage in patients with a fault in a gene known as C9orf72. This faulty gene is known to cause Amyotrophic Lateral Sclerosis (ALS; also known as motor neuron disease or MND) and a linked condition called Frontotemporal Dementia (FTD). To carry out the study, we need to obtain blood and other tissue samples from carriers of the faulty version of the C9orf72 gene who have ALS or FTD, as well as their family members who do not have ALS or FTD symptoms. We also need to study a group of people with ALS or FTD who do not have this faulty gene, in order to compare results. Finding out why patients carrying the faulty C9orf72 gene develop ALS and FTD may help us in the future to find treatments to help prevent these conditions.

The C9orf72 fault (also known as a mutation) is the most common genetic cause of ALS and FTD and is present from birth in patients who carry it. By studying samples such as blood, skin, and spinal fluid from patients and relatives, we can gain an understanding of the processes that are at work in this inherited form of ALS and FTD. Samples from blood and the fluid that surrounds the brain as well as brain imaging and electrical nerve tests, can

help us study the disease at a molecular level. This will help us to increase our understanding of the underlying biology that is relevant to the faulty gene and eventually help us to better monitor responses to treatments during future research studies. Eventually, all of this knowledge may contribute towards the development of new treatments for patients with ALS and FTD who have the faulty C9orf72 gene and ALS and FTD in general.

The study will also lead to the curation of a national register of C9orf72 patients, and their asymptomatic family members, for possible inclusion in future clinical trials or other academic studies (subject to further consent).

Why have I been invited to participate?

You have been asked to participate in this study as you were nominated as a "study partner" by someone taking part in the study. They will either have a diagnosis of ALS or frontotemporal dementia, be taking part as a family member of someone with the C9orf72 genetic abnormality, or may be a healthy control participant. You will be asked to provide additional information about the person taking part. If you wish to contribute your own information, scans and samples to the study, you will be provided with further information about participating.

Do I have to take part?

No. It is up to you to decide whether or not to take part. If you do take part, you will be given this information sheet to keep and you will be asked to sign an online or paper consent form. You are still free to withdraw at any time and without giving a reason.

We will ask you if you would be willing to be contacted about other studies in the future. You can decline to be contacted and this will not impact on your ability to participate in the current study or your legal rights or any future clinical care.

What do I have to do to take part?

We will ask you to provide information (in person, by video or over the telephone) on your friend’s or relative’s general health and medical care as well as information about your own wellbeing. We will ask you and your friend or relative to sign a consent form before entering the study.

We will contact you via telephone, video call or mail, or may ask you to come to the John Radcliffe Hospital in Oxford with the person you are providing information for, for research visits where they undergo a variety of clinical and cognitive assessments and tests. These visits may take place at 3-24 month intervals, until the study is complete, the person you are accompanying no longer wishes to, or is unable to participate, or the research team decide to withdraw him/her from the study.

At each visit:

You will be asked a series of questions regarding their cognition (e.g. memory and attention) and behaviour using a questionnaire which is in regular use for this purpose.

We will administer this questionnaire while the person you are accompanying is undertaking another part of the study.

Your part of the questionnaires will take around 30 minutes, after which you would be free to leave.

What are the possible benefits of taking part?

There is no intended clinical benefit to you for taking part in this study, but we hope that the information we obtain will eventually help to improve the diagnosis and treatment of people who carry the C9orf72 gene mutation.

Will my taking part in the study be kept confidential?

Yes. All the information about your participation in this study will be kept confidential.

Responsible members of the University of Oxford [and the relevant NHS Trust(s)] may be given access to data for monitoring and/or audit of the study to ensure that the research is complying with applicable regulations. All will have a duty of confidentiality to you as a research participant and nothing that could reveal your identity will be disclosed outside the research site. We will ask you to consent to allow these people access to the information collected about you in the course of the study. All people looking at your records and the procedures for handling, processing, storage and destruction of your data are compliant with United Kingdom General Data Protection Regulation (UK GDPR) and the Data Protection Act 2018.

All data will be stored securely by the research unit for at least 20 years after the study has been completed.

Will I be reimbursed for taking part?

Reasonable travel expenses for any visits you make to Oxford for this study on production of receipts, or a mileage and parking allowance provided, as appropriate. There is no other payment for taking part in this study.

What will happen to my data?

We will be using information from you and your medical records in order to undertake this study. United Kingdom Data protection regulation requires that we state the legal basis for processing information about you. Research is a task that we perform in the public interest. The University of Oxford, based in the United Kingdom as Sponsor, is the data controller.

This means that we, as University of Oxford researchers, are responsible for looking after your information and using it properly. We will use the minimum personally-identifiable information possible. We will keep identifiable information about you for three years after the study has finished. We will store the research data and any research documents with personal information, such as consent forms, securely at the University of Oxford for up to 20 years after the end of the study as part of the research record.

The local study team will use your initials, date of birth, and contact details, to organise follow-up visits (if applicable) and to report any unexpected findings with clear implications for your current health (please see the section entitled "What if you found an unexpected abnormality?" below). They will keep identifiable information about you from this study for three years after the study has finished.

UK Data protection regulation provides you with control over your personal data and how it is used. When you agree to your information being used in research, however, some of those rights may be limited in order for the research to be reliable and accurate. Further information about your rights with respect to your personal data is available at https://compliance.web.ox.ac.uk/individual-rights

You can find out more about how we use your information by contacting the research team at acorn@ndcn.ox.ac.uk.

What will happen to the results of this study?

The results of this research may be presented at scientific meetings in the UK and overseas. It will not be possible to identify you from any of the data that will be presented. The data from the study may also be published in a medical journal. You will not be identified in any report or publication. The results of this study may lead to the development of patents and/or to commercial benefits for sponsors and researchers. A patent is a right to the exclusive use of an invention, such as a new test or new drug, for a fixed period of time. You would not be entitled to receive any personal or financial benefit from this.

What if there is a problem?

If you wish to complain about any aspect of the way in which you have been approached or treated, or how your information is handled during the course of this study, you should contact Dr Alexander Thompson on 01865 228371 or by email alexander.thompson@ndcn.ox.ac.uk or you may contact the University of Oxford Research Governance, Ethics & Assurance(RGEA) office on 01865 616480, or the director of RGEA, email RGEA.Complaints@admin.ox.ac.uk

The Patient Advisory Liaison Service (PALS) is a confidential NHS service that can provide you with support for any complaints or queries you may have regarding the care you receive as an NHS patient. PALS is unable to provide information about this research study. If you wish to contact the PALS team at the John Radcliffe Hospital, please phone 01865 221473 or

email PALS@ouh.nhs.uk.

The University of Oxford, as Sponsor, has appropriate insurance in place in the unlikely event that you suffer any harm as a direct consequence of your participation in this study.

Who is organising and funding the study?

This research study is organised by the University of Oxford.

The funding for this study comes from the National Institute for Health Research – Oxford Biomedical Research Centre, and the Alan Davidson Foundation in a charitable gift to the Oxford MND Care and Research Centre.

Who has reviewed the study?

All research in the NHS is looked at by an independent group of people, called a Research Ethics Committee, to protect participants' interests. This study has been reviewed and given favourable opinion by the London-Surrey Research Ethics Committee.

Participation in future research:

Independent from the current study, and if you consent, we may offer you the opportunity to participate in other studies. These studies will be explained separately, and you would give consent for them specifically. Agreeing to be contacted about future research does not oblige you to take part. You can decline to be contacted and this will not impact on your ability to participate in the current study or your future clinical care. Your contact details would be held separately from the study data in a password protected database, on a computer in the Nuffield Department of Clinical Neurosciences. You have the option of removing your contact details at any time by contacting the research team.

Further information and contact details:

Please contact the ACORN study team at the Oxford MND Care and Research Centre at acorn@ndcn.ox.ac.uk

Thank you for reading this information sheet and considering taking part in this study. If you decide to take part you will be given a copy of this information sheet and a PDF or paper copy of the signed consent form.