MRI in Pompe study

Patient information Sheet for Adults – Patients with Pompe

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Sponsor: The Newcastle Upon Tyne NHS Hospitals Foundation Trust

Chief Investigator: Professor Jordi Diaz Manera

Introduction

You are being asked to take part in this research study because you have Late Onset Pompe Disease.

Clinical research studies are research studies which include only people who freely choose to take part. Please take your time to read this information carefully. It is important that you read and understand the contents of this participant information sheet.

This participant information sheet gives you important information about the study to help you decide if you want to participate. It describes the purpose of this study, the study procedures, the possible risks and benefits, the amount of time required for the study, and provides information about your rights as a study participant. Your consent is required for participation.

If you are unsure of anything within this participant information sheet, or if you have any questions or queries, please discuss this with your study team. You can also discuss this participant information sheet with family or friends or your primary care or specialist doctor before making your decision to take part in this study. If you decide you would like to take part in this study, you will be asked to complete the informed consent form.

Your permission to take part in this study is voluntary. You are free to say yes or no. If you do not want to participate, your regular medical care and legal rights will not be affected. Even if you join this study, you may stop your participation at any time and your regular medical care and legal rights will not be affected.

Please tell the study doctor or study staff if you are taking part in another research study.

Why have I been asked to take part?

You have been asked to participate in this study because you have Pompe disease.

Why is this study being done?

The purpose of this study is to study if glycogen, the sugar that is being accumulated in your muscles, can be detected and quantified using a new imaging tool known as muscle magnetic resonance, specifically using carbon spectroscopy. If so, this imaging technique could be useful for 1) diagnosing patients with Pompe disease, 2) monitoring disease progression, 3) understanding if increases in glycogen in the muscle worsens muscle function and could be therefore used as an indicator to start treatment and; 4) to monitor response to enzymatic replacement therapy and other potential drugs in the future aiming to reduce the amount of glycogen in the muscles.

How many people will be involved in the study?

This study will include 20 participants – 10 people with Pompe and 10 people who do not have Pompe, but whose gender and age will be matched to the people with Pompe.

Do I have to agree to take part?

Participation in the study is voluntary, you can decide whether or not you want to take part. If you do agree, you will be given this information to keep and will be asked to sign a form which gives your consent to take part. If you do decide to take part and then you change your mind, you are free to withdraw at any time and without giving a reason. This will not affect the standard of care you receive.

What would taking part involve? What will happen if I agree to take part?

If you agree to take part in this study, you will be asked to sign a separate Informed Consent Form. We will answer any of your questions about the study before asking you to sign the consent form.

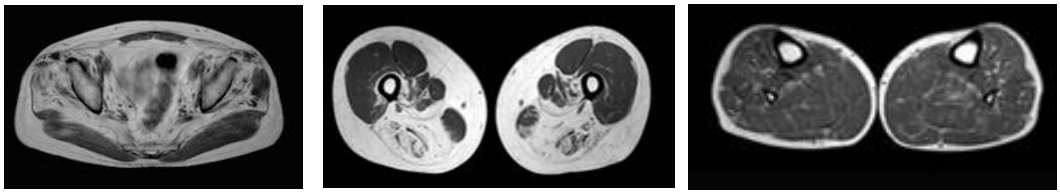
The study involves two visits, one at baseline and another visit one year after.

Both visits will include clinical assessment, collection of blood samples, a series of muscle function tests and collection of patient reported outcome measures, performed at the Newcastle Clinical Research Facility. Muscle function tests in this study measure how you walk, the distance you can walk in 10 seconds, how you move in general (for example doing squats or standing up from a chair). The estimate time needed for the muscle test is 30 minutes per participant. Both visits will also include Magnetic Resonance Imaging (MRI) scans, which will be performed at the Newcastle Magnetic Resonance Centre.

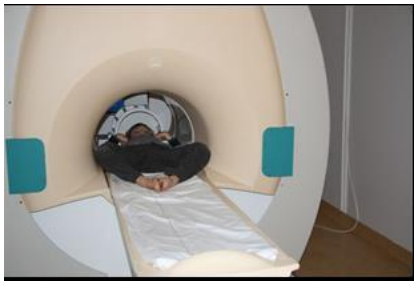
We will collect approximately 2 tablespoons of blood. Samples will be collected at the MRC Neuromuscular Biobank. Samples will not be analysed now but they are going to be stored for future studies. We plan to quantify different molecules that could be related with disease progression and muscle MRI findings such as microRNAs or cytokines. After that samples will be stored for future use in case there are new molecules described that we would like to check in the samples of these patients.

What is an MRI scan?

MRI stands for Magnetic Resonance Imaging. This technique uses magnetism and radio frequency waves to collect information about the part of the body being examined, in your case, the MRI scan will be performed only on your thigh. The radio waves are bounced back to the scanner by your body and a computer within the scanner uses this information to produce images. Examples of such images are shown below.



The MRI scanner looks like a tunnel with open ends and you will lie on a table that slowly slides into the tunnel with the feet first before the scan begins.



To obtain proper images, it is important that you lie very still. As it is much easier to lie still when you are comfortable, extra time will be taken to increase your comfort as much as possible. You will lie on your back on the scanner bed with your legs slightly elevated on a pillow. A helmet will be placed over your head with a mirror that allows you to look out of the tunnel. You will be wearing headphones to protect your hearing from the noise the scanner makes. The same headphones are used for communication with the researcher and music of your choice can be played over them to increase the comfort. Some people fall asleep during an MRI scan, which is not a problem.

Please note that a hoist will be available to help you lie on the bed of the MRI scanner if needed.

How long will I be in the study?

You will be asked to attend two visits over a period of one year.

What are the potential risks or discomforts?

The MRI is a safe procedure, which gives researchers detailed information about the area being examined. It does not use x-rays and so it does not involve any risk from radiation exposure. The scanner is quite noisy and some people feel claustrophobic during the examination and others find it difficult to lie still. If you find the scan too uncomfortable it can quickly be stopped.

An MRI does not have any impact on the development of a foetus, however we prefer not to include pregnant women in the study to avoid any potential complications. Muscle MRI can also be performed in women that are breastfeeding without provoking any damage.

During the collection of blood samples, you may experience pain and/or bruising at the place on your arm where blood is taken. If you would like, an anaesthetic cream can be applied to make the area numb and reduce discomfort. Rarely, a clot may form and infections occur where the blood is taken, or you may faint. Study staff will be able to help if this happens.

What are the possible benefits of taking part?

There will be no immediate clinical benefit to you as a result of your participation in this study, but we hope that the information obtained from this study will help us to know if glycogen can be identified and quantified on your skeletal muscles using carbon spectroscopy. This will help in the diagnosis

and monitor of patients with Pompe disease in several ways. For example, this imaging technique could be useful for 1) diagnosing patients with Pompe disease, 2) monitoring disease progression, 3) understanding if increases in glycogen in the muscle worsens muscle function and could be therefore used as an indicator start treatment and; 4) to monitor response to enzymatic replacement therapy and other potential drugs in the future aiming to reduce the amount of glycogen in the muscles.

Is there anything I need to do before the MRI?

When consenting to your MRI, please tell your researcher if you have:

- had any recent surgery;
- any surgical clips;
- a previous history of metal fragments in the eyes;
- any history of asthma
- any type of dental braces

What happens if something abnormal is discovered?

When having a scan, there is always a small possibility that an abnormality could be observed on the images of which you and your doctors are unaware. The MRI scans we collect will be reviewed by a radiologists in the Newcastle upon Tyne Hospitals NHS Foundation Trust to look for any such findings.

It is important to recognise that the MRI scans are not being taken for diagnostic purposes and so there is no guarantee that the scans would be of the right kind to detect any abnormality which may be present.

Should the radiologist suspect anything abnormal on your scans they will inform the study Principal Investigator who will contact your clinical care team or GP in order to make recommendations about any further investigations which might be appropriate for them to arrange.

Who is organising and funding this study?

This study is funded by a grant received from Sanofi- Genzyme and they do not have any role in the design of the study. They will receive a report of the study with anonymized aggregated data showing the results. The Newcastle Upon Tyne NHS Hospitals Foundation Trust are the study Sponsor. The study will be conducted in conjunction with Newcastle University and The Newcastle Upon Tyne NHS Hospitals Foundation Trust.

What if relevant new information becomes available?

Sometimes during a study new information becomes available. If this happens your study doctor will tell you about it and discuss with you whether you want to continue. If you decide to withdraw your study doctor will ensure that your care will continue. If you decide to continue, you may be asked to sign a new consent form.

What will happen if I don't want to carry on with the study?

You can leave the study at any time without giving a reason and this will not affect the care that you receives now or in the future. If you decide that you would like to withdraw completely from the study, we would like to retain any data collected up to the point of withdrawal, for our research.

How will my information be kept confidential?

All the information that we will collect about you during the course of the study will be kept strictly confidential. Information you give us will be stored in a secure database. You will not be able to be identified in any ensuing reports or publications.

The database linking unique sample study numbers to personal details will only be accessed by authorised members of the research team. Information we have stored about you will not be used or made available for any purpose other than for research and improvements in health care.

Your name, family name or any other personal identifiers will not be in any report or publication, including information about the study or the data gathered. Any research data generated and made available to others for further research will have your personal identity removed.

We will need to use information from your medical records for this research project.

This information will include your name/ NHS number/ date of birth/ gender / ethnicity and if applicable any of the following: diagnosis and management of Pompe /functional assessments / age at assessment / medications / any relevant clinical, genetic or biochemical history and health information. People will use this information to do the research or to check your records to make sure that 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 but you can request for your data to be removed from the database. We will write our reports in a way that no one can work out that you took part in the study.

What are my choices about how my information is going to be used?

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 for the research to be reliable. This means that we will not 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/
- our leaflet available from www.hra.nhs.uk/patientdataandresearch
- by asking one of the research team

What will happen to the results of this study?

The results from the research study may be used to write articles to be submitted for publication in scientific or medical journals. It is also likely that the results of this study will be presented at meetings and conferences. The results will also be sent to the organisation that funded the research, in a form of a report. However, your identity will not be revealed in any reports or publications. The study results will not be directly provided to study participants.

How have patients and the public been involved in this study?

This protocol has been reviewed by Allan Muir, Chair of the Pompe Support Association in UK and by the Association for Glycogen Storage Disease in UK. They have suggested some changes to the protocol that have been amended.

Who has reviewed this study?

All research studies are reviewed by an independent Research Ethics Committee to protect your safety, rights, wellbeing and dignity. This study has been reviewed and approved by [INSERT NAME OF THE ETHICS COMMITTEE]. The reference for this study is: [INSERT REFERENCE NUMBER].

Will I get paid to take part in the study?

You will not receive payment for taking part in this study. Travelling expenses will be reimbursed for you to attend study visits.

Informing General Practitioner (GP) / other healthcare practitioner

With your permission, we will let you GP, and other healthcare professionals involved in your care, know that you are taking part in the study.

What will happen to the samples I give?

The samples collected will be stored at the Newcastle MRC Centre Biobank for Rare and Neuromuscular Diseases until they will be used for research. The blood samples collected are planned to be used for future studies aiming to identify biomarkers of disease progression correlating with results of muscle function test or MRI. You can opt out at any time and you can request for your sample to be destroyed.

What happens if I get hurt taking part in this study?

There are no special compensation arrangements in the event that something goes wrong and you are harmed during the research study. Newcastle University has insurance cover for the design of the study. NHS bodies have insurance for clinical negligence for people under their care. If you are harmed and this is due to someone's negligence, you may have grounds for legal action for compensation against Newcastle University or the NHS (their employer). The normal NHS complaints mechanisms will still be available to you. Please contact a member of study staff at the site if any injuries are sustained during this study that are related to the research.

What if something goes wrong?

If you have a concern about any aspect of this study, you should ask to speak to the researchers who will do their best to answer your questions

Principal Investigator: Professor Jordi Diaz-Manera

Email: Jordi.Diaz-Manera@newcastle.ac.uk

Telephone: 0191 241 8602

John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, International Centre for Life, Newcastle upon Tyne, NE1 3BZ, United Kingdom

If you prefer to raise your concerns with someone not involved in your care, you can contact the Patient Advise and Liaison Service (PALS). This service is confidential and can be contacted on Freephone: 0800 032 0202

Email: northoftynepals@nhct.nhs.uk

Alternatively, if you wish to make a formal complaint you can contact the Patient Relations Department through any of the details below:

Telephone: 0191 223 1382 or 0191 223 1454

Email: patient.relations@nuth.nhs.uk

Address: Patient Relations Department, The Newcastle upon Tyne Hospitals NHS Foundation Trust, The Freeman Hospital, Newcastle upon Tyne, NE7 7DN

If you wish to raise a complaint on how we have handled your personal data, you can contact our Data Protection Officer (see contact details below) who will investigate the matter. If you are not satisfied with our response or believe we are processing your personal data in a way that is not lawful, you can complain to the Information Commissioner's Office (ICO). Further details of how to do this can be found on the ICO website:

https://ico.org.uk/for-organisations/data-protection-reform/overview-of-the-gdpr/individuals-rights/

What if I have any more questions?

If you have any questions, please contact Prof Jordi Diaz-Manera

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