MRI in Pompe disease study

Muscle MRI in Pompe disease

Patient Information Sheet for 12+ Patients with Pompe

Version: 3.0 | 28.07.2025

Sponsor:

The Newcastle Upon Tyne NHS Hospitals Foundation Trust

Chief Investigator:

Professor Jordi Diaz Manera

Introduction

We are inviting you to take part in a research study. Before you decide whether or not you want to take part, it is important that you know why the research is being done and what it will involve. Please read the following information carefully, and talk about it with your parents/guardian, friends or your doctor, if you want to. If there is anything that you are not sure about, or if you want to ask anything, you can at any time. Take time to decide whether or not you would like to take part.

If you do decide that you want to take part, you are free to change your mind at any time without telling us why and it will not affect any of your normal care.

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 includes up to 25 participants, children and adults:

- 15 people with Pompe and,
- 10 people who do not have Pompe, but whose gender and age will be matched to 10 of the people with Pompe.

You belong to the cohort of 5 patients in middle stages of the disease progression.

Do I have to agree to take part?

No, it is up to you if you want to take part. If you say yes, you will be given this sheet to keep and be asked to sign a form to say that you want to take part. If you say yes and then you change your mind, that is fine, and your care will carry on as normal.

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

For this study, you and your parent/guardian will need to attend two visits, one at the beginning of the study and one after one year.

At both visits, a researcher will ask you and your parent/guardian some questions about you and will look at your records to see the results of tests that you have had on different parts of your body.

We will then perform an MRI (Magnetic Resonance Imaging) clinical assessment, taking blood samples (approximately 2 tablespoons of blood) and a urine sample, a series of muscle function tests and we will then ask you and your parents/guardian some questions about how you are doing and any medicines you are taking. For these visits, you will come to the Newcastle Magnetic Resonance Centre to perform the MRI, and to the Newcastle Clinical Research Facility at the Royal Victoria Infirmary, for the blood and urine samples and the physio assessment.

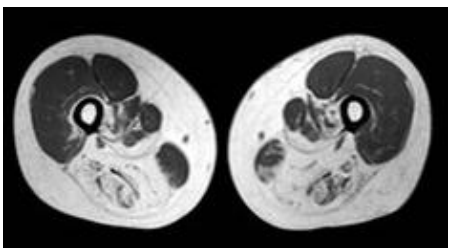
The MRI will take around 1 hour, and it will be done just on your lower limbs. This means that your head will be out of the scanner, and you won't feel claustrophobic.

Muscle function tests 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) and will take about 60 minutes per participant.

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.

What is an MRI scan?

MRI stands for Magnetic Resonance Imaging. You will lie on a table, and the machine makes pictures of your leg muscles, like the pictures 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 get good pictures it is important that you lie very still. We will spend time making sure that you are comfortable. It does not hurt to have a scan. You will be wearing headphones to protect your ears because the scanner makes noise. The researcher can talk to you through the headphones, and you can also bring your favourite music to listen to. There will be several pictures, each taking about five minutes. The pictures will take about 1 hour in total to take. Your parent / guardian can come with you into the MRI room.
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. If you are already receiving enzyme replacement therapy (ERT), you may also be asked to attend an additional visit approximately one week after your first visit, to study how your treatment affects the glycogen content in your thigh muscles. We will explain clearly to you and your parent or guardian, before you decide to take part, if you will need to come for one or two visits at the beginning of the study.

Are any of the tests risky?

The scan is safe, but it is quite noisy and it does require you to stay still. To reduce these difficulties, you will be able to listen to music and you can bring some along with you. It will be much easier to lie still if you lie comfortably. The researchers have a lot of experience increasing comfort levels using various pillows and soft straps. Please do not hesitate to tell them which ones you like. If you find the scan too uncomfortable it can quickly be stopped.

What are the possible benefits of taking part?

There will be no benefits for yourself in you taking part but we hope that the new knowledge from this study will help other people with Pompe Disease in the future.

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

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. The Newcastle Upon Tyne NHS Hospitals Foundation Trust is 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 receive 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?

Only the trained staff working on the study will know your name and have access to your medical records. All forms used will have a code on them instead of your name so no one will know who you are.
We will keep all information about you safe and secure.

What will happen to the results of this study?

The results from the study may be presented at meetings and be used to write articles to scientific or medical magazines. Your name will never be used on these occasions.

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 and your parent/guardian to attend study visits.

Informing General Practitioner (GP) / other healthcare practitioner

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

What if something goes wrong?

If you or your parent/guardian 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
- Address: The 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 or your parent or carer are unhappy or worried about anything in the study, you can talk to someone in the research team.

If you want to speak to someone not involved in your care, you can contact the Patient Advice and Liaison Service (PALS). This is a friendly and private service that can help you:

- Freephone: 0800 032 0202
- Email: northoftynepals@nhct.nhs.uk

If you want to make a formal complaint, your parent or carer can contact:

- Patient Relations Department
- Phone: 0191 223 1382 or 0191 223 1454
- Email: patient.relations@nuth.nhs.uk
- Address: Patient Relations, Freeman Hospital, Newcastle upon Tyne, NE7 7DN

If you or your parent/carer are worried about how we keep your personal information safe, you can contact our Data Protection Officer:

- Name: Julia Scott
- Email: julia.scott12@nhs.net
- Phone: 0191 223 1862

You can also find out how to make a complaint about your personal data by visiting the Information Commissioner's Office (ICO): www.ico.org.uk/make-a-complaint/data-protection-complaints

What if I have any more questions?

If you want to ask anything you can ask your parents / guardian to get in touch with the researchers at any time. If you have any questions, please contact Prof Jordi Diaz-Manera:

- Email: Jordi.Diaz-Manera@newcastle.ac.uk
- Telephone: 0191 241 8602
- Address: John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute University of Newcastle upon Tyne, International Centre for Life, Central Parkway, Newcastle upon Tyne, NE1 3BZ, United Kingdom