MRI in Pompe study

Patient Information Sheet for 12+ – HEALTHY CONTROLS

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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, 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 as a healthy volunteer because your gender and age is matched to someone with Pompe Disease who is taking part. Pompe disease is a genetic disease and patients with this condition develop muscle weakness that can lead to permanent disability.

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, children and adults – 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?

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 parents/guardian will need to attend only one visit.

At the visit, a researcher will ask you and your parents/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.

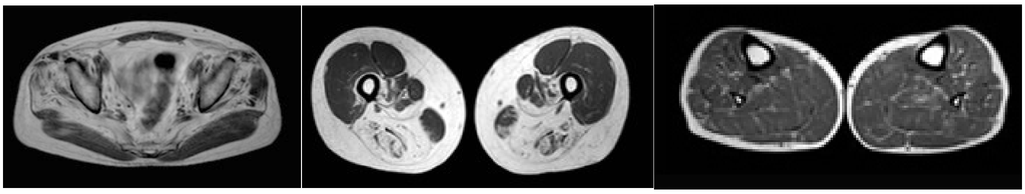
We will then perform an MRI (Magnetic Resonance Imaging) clinical assessment, taking blood samples (approximately 2 tablespoons of blood), and a series of muscle function tests and we will then ask you and your parents/guardian some questions about how you are doing and any medicines you are taking. For this visit, 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 samples and a physio assessment.

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.

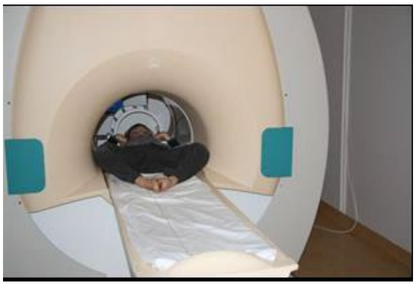
If you agree to take part in this study, you will be asked to sign a separate Informed Consent Form. We will answer any of your questions about the study before asking you to sign the consent form.

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. The MRI looks like a tunnel that is open at both ends and you will lie on your back on the scanner bed. It does not hurt to have a scan. A helmet will be put on your head with a mirror so you can look out of the tunnel. You will be wearing headphones to protect your ears because the scanner makes noise. The helmet is big enough to fit over the headphones. 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 45 minutes in total to take. Your parent/guardian can come with you into the MRI room.

How long will I be in the study?

You will be asked to attend only one visit.

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 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 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?

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 in 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.

What if something goes wrong?

If you or your parents/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

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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

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