PhenoDM1 - Myotonic Dystrophy type 1 (DM1) deep phenotyping to improve delivery of personalised medicine and assist in the planning, design and recruitment of clinical trials.

Patient Information Sheet

We would like to invite you to participate in a research project called PHENODM1. Before you decide whether or not to participate, we need to be sure that you understand why we are doing the study, and what it would involve if you agreed to take part. Please take time to read this information carefully. We will do our best to explain and provide any further information you may ask for now or later. You do not have to make an immediate decision to participate if you are not ready. This study may have no direct benefit to you, but will provide important information for possible future trials.

Why have I been invited to take part in this study?
You have been contacted because you have a rare neuromuscular disease, myotonic dystrophy type 1.

What is the purpose of this study?
Myotonic dystrophy type 1 is a progressive disease with varied symptoms which often include progressive muscle weakness, daytime sleepiness and fatigue. There are no known medicines to treat the major symptoms of myotonic dystrophy type 1 with the exception of the drug Modafinil, which is sometimes used to help control excessive daytime sleepiness.

This study will collect lots of information about people with myotonic dystrophy type 1 and record how things change or stay the same over the course of a year. Research into myotonic dystrophy is moving very quickly and having a well-described group of patients will make it easier and faster for treatments being developed in the laboratory to be tested in clinical trials.

Who can participate?
In order to take part you must be over 18 and have been diagnosed with myotonic dystrophy type 1 through genetic testing. You should also be able to walk independently and be able to provide informed consent.

Do I have to take part?
No. It is entirely up to you whether or not to take part. Participation in this study is entirely voluntary and you are free to withdraw from the study at any time without having to give a reason. This will not affect your future medical care or your relationship with medical or nursing staff looking after
you. If you, your study doctor, or one of your clinicians decides you should withdraw from the study, we would like your permission to keep and analyse the data already collected.

What will I have to do?
You will be asked to visit the study site twice approximately 12 months apart. Each visit will last approximately 4 hours. During both visits you will be asked to complete the same assessments, these assessments will measure lots of different aspects of myotonic dystrophy and how it affects you.

At the visits we will:

- Ask you to complete some questionnaires about your myotonic dystrophy and how it affects you, this will include questions about pain, fatigue and quality of life.
- Check your heart and lung function by performing the following tests;
  - Electrocardiogram (ECG), this routine test takes just a few minutes and involves you lying on an examining table with a number of electrodes attaches to your arms and chest.
  - Spirometry/Force vital capacity (FVC), this routine test involves you breathing into a tube as hard as possible three times.
- Ask you to complete the six minute walk test
  - This involves walking up and down a corridor for 6 minutes; we measure how far you can walk and how many times you need to rest and ask you to rate your perception of excretion during the walk.
- Ask you to complete the ten minute walk test
  - This is the same as the six minute walk test but lasts ten minutes.
- Ask you to complete a timed up and go test
  - This test is a measure of the time it takes you to get up from a seating position walk 3 metres (10 feet) and return to the seating position.
- Check your age, height and weight.
- Ask you to give a blood sample (30mls, or about 2 tablespoons)
- Ask you to give us permission to access any previous DNA samples that may have been stored after your diagnosis. This will be used for research purposes which may not have a direct impact on your care. All samples and data associated with them will be anonymised.

What happens to my normal care? We will not alter your medication or change any other treatment you receive in any way. You should continue to attend all medical appointments as normal. However, if there are any changes to the medication you are taking you should inform the study team. Your GP will be informed of your participation, if you consent to this, and will be asked to contact the trial team to discuss any changes to your medication.

Expenses and payments
You will not be paid to take part in this study but your travel expenses will reimbursed unless this is a routine clinic appointment.

What are the possible benefits of taking part in the study?
You will be monitored closely during the study by the study team. The tests we do will give us (and you) information about your general well-being. If any of these investigations reveal any new abnormality we will let you, your GP, and clinicians know. The study may not immediately benefit you, but in collecting lots of information about people with myotonic dystrophy type 1 may improve the quality of care and management for people in the future.
What are the possible disadvantages and risks in taking part?
The additional visits to the study site may be inconvenient and having blood samples taken may be uncomfortable for some people. You will be asked to complete questionnaires relating to different aspects of your symptoms for example; pain, fatigue and general well-being. The research nurse or therapist will be able to assist you to complete the questionnaires.

Will the details about me be kept confidential?
Yes. Best ethical and legal practice will be followed to ensure that all information collected will be handled in confidence and in accordance with the Data Protection Act 1998. The database linking unique sample study numbers to personal details will only be accessed by authorised members of your Clinical Care Study team and individuals from the Rare Diseases – Translational Research Collaboration. You will not be identified personally in any report or publication, including information about the RD-TRC studies which will be released on the Internet.

Who is funding this research?
The study is being led by Professor Hanns Lochmuller at University of Newcastle and Newcastle upon Tyne Hospital NHS Foundation Trust. The study is funded by the National Institute of Health Research (NIHR).

Who has reviewed the study?
The Newcastle and North Tyneside 1 Ethics Committee reference NE/15/0178 which has responsibility for scrutinising proposals for medical research on humans, has examined the proposal and has raised no objections from the point of view of medical ethics.

What if something goes wrong?
If you have a complaint about your participation in the study you should first talk to a researcher involved in your care. You can ask to speak to a senior member of the research team or the Complaints Officer for Newcastle upon Tyne Hospitals NHS Foundation Trust at the Patient Relations Department on 0191 223 1382. In the event that something goes wrong and you are harmed during the study there are no special compensation arrangements. If you are harmed and this is due to someone’s negligence then you may have grounds for a legal action for compensation against Newcastle upon Tyne Hospital NHS Foundation Trust but you may have to pay your legal costs. The normal National Health Service complaints mechanisms will still be available to you (if appropriate.)

If you feel that you have been treated unfairly throughout the research, or would like to comment on the conduct of any aspect of this please contact:

Contact details
For additional information contact: Libby Wood, clinical research associate on 0191 241 8640 or email Elizabeth.wood2@newcastle.ac.uk

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If during the study you become unwell or are concerned, contact NHS Direct.. If you are unwell and need urgent advice or assistance do not delay in seeking further advice or treatment as usual through the NHS services.

Thank you for reading this information sheet and considering taking part in this study.