



UX001-CL401



International HIBM Registry
Patient Information and Informed consent

Project Name: Hereditary Inclusion Body Myopathy-Patient Monitoring Program (HIBM-PMP): A Registry and Prospective Observational Natural History Study to assess HIBM Disease

Protocol Number: UX001-CL401
Date/Version: 23 May 2013/2

You are being asked to take part in the registry. Before you agree to take part in this study, it is important that you understand what this research study is all about and what will be done with the information that you provide. This form contains the answers to questions that you might have. However, if after reading this form you have additional questions relating to the study, you can contact either the registry coordinator or authorised staff prior to signing the consent form. Contact information is included at the end of this document.

Patient information and consent form were reviewed by two members of patient organisation Neuromuscular disease foundation.

Summary

Hereditary Inclusion Body Myopathy (HIBM), also known as Nonaka disease, GNE myopathy or distal myopathy with rimmed vacuoles is a rare muscle disease. You are being asked to participate in the international database (registry) for patients with HIBM disease. Registering with the database means that we will ask you to fill in questionnaires to provide details of your condition, general medical history, medications you are taking, quality of your life and ability to move. This will take approximately 1.5 hours to complete all questions at the first time. You can complete as many questions as you want and return to your profile to complete the rest of the questions at a later time.

We will contact you in 6 month, 12 month and yearly after to ask you to update your profile by completing a short form. If you agree we will contact you to inform about advances in HIBM research, studies and trials in HIBM.

Your agreement to participate in the registry is voluntary and you can withdraw from the registry at any point without giving any reason. Regardless of your participation in the registry your medical care will not be affected in any way. Your consent to participate in this study will be taken electronically. The registry will be stored online and data can be accessed from the UK and USA by regulatory bodies (FDA and DHHS) and authorised researchers.

Registering for the database over the internet may pose some risk to data security; however, significant efforts are put in place to ensure this does not happen. The registry documentation and policy has been reviewed and is compliant with best practice and national legislation (Caldicott approval) was granted in the UK). All information we receive from you will be treated confidentially.

Who is doing this study?

TREAT-NMD (Newcastle University), a steering committee, consisting of HIBM medical experts and patient advocates, along with Ultragenyx Pharmaceutical is doing this research. TREAT-NMD has extensive experience in similar rare muscle disease registries. For more information please see: www.treat-nmd.eu

What is a patient registry and why do we want to create one?

When a clinical study or trial is being planned, it is very important that patients suitable for that trial can be found and contacted quickly. A registry is like a bridge connecting patients and families with doctors and researchers who are trying to understand and treat the disease by making sure that patients' details are all collected in a single database or "registry". TREAT-NMD network and Ultragenyx Pharmaceutical are creating this international registry for people from different countries who have HIBM.

Whose data are we collecting in this registry?

This registry is for patients over 18 years affected by HIBM. This is a muscle weakening disorder uncommon in the general population. You are being asked to be in this registry and to fill this form, because you have this condition. You may ask your doctor for help in answering some of the questions. Although a number of patients have been identified worldwide, there has been only one national registry launched for HIBM patients in Japan. Our aim is to create an international registry to better understand the global patient population and facilitate access of HIBM patients worldwide.

What information am I required to provide?

You will be asked questions about your history of HIBM: when, where and how you were diagnosed, what treatments you may have taken to help manage your condition, and whether any family members also have HIBM. You will be asked to share results of your DNA analysis, your general medical history and any previous or current medications you may be taking. You will complete a series of questionnaires that will ask about your quality of life, how well you can move around and care for yourself. Based on the information you provide, staff in charge of the registry might need to gain access to your medical records to obtain information necessary to the project (for example we may need to ask your geneticist or a doctor for a copy of your genetic report and also information on your muscle biopsy).

What happens if I do not want to answer some of the question or do not know the answer to a question on the form?

You can complete as many questions as you want at a time. You can ask your doctor to help in answering some questions. You can always return to your profile and complete the rest of the questions at a later time. We encourage participants to complete as many questions as possible because it will help to get a better understanding of HIBM.

Who will have access to my information?

The information that you enter will be entered into an international registry which is supervised by TREAT-NMD. Your data will be stored securely and no unauthorised people will be able to gain any information about you. Only researchers, who have been

approved by their local ethics committee and by the TREAT-NMD, steering committee, and Ultragenyx Pharmaceutical Inc, are allowed access to the registry anonymous data.

Your information (apart from names and contact details, which will not be disclosed) may also be shared with the following individuals or groups so that they can carry out their duties related to the study:

- The study sponsor, Ultragenyx Pharmaceutical, Inc. will have an access to de-identified data in order to analyse results and plan future clinical studies and trials.
- Ultragenyx collaborator (Summit Analytical) will have an access to the database in order to provide database maintenance and ensure its proper functioning and to help analysing the data.

Those who receive your information may share it if they are required to do so by law:

- UK and USA regulatory bodies (such as FDA and DHHS) may review or obtain your information (including your identity and name) in order to confirm the accuracy of the research data.
- An independent committee of physicians and experts that will review safety information from this study.
- Individuals or businesses such as data storage companies or IT companies outside the research site that provide services
- If required by law, regulatory agencies from other countries may review provided information for quality assurance or quality control purpose. .

In the registry, your data will only be identified by an anonymous code, not by your name.

This means that when researchers search the registry they will not be able to find out your personal information (name, address, etc.), but only the information they need to know about your condition that will help them decide whether you might be suitable for the study or trial. If they think that you meet the criteria for their study they will contact the person in charge of the registry. Staff working for the registry will “de-code” the data to find out the personal details and will contact you to give you information about the trial or about any other issues relevant to your condition. They will not give your name or personal information to the researchers and will not be able to contact you directly unless authorised by you. If you are interested in the information that you receive about a particular clinical study/trial, you will be given information about how you can contact the researchers running it. If you decide to take part in the study/trial, you will need to review and sign a separate consent form. Signing up for the registry does not entitle for participation in studies or trails.

How can I update my data if something has changed?

If you consent to be a part of this study, you will be asked to provide your information at baseline, 6 and 12 months, then yearly thereafter for up to 15 years. We will send you emails reminding you to update your information. We also ask you to inform us if there are any major changes in your details that might occur in the period between updates, for example change of address or loss of ambulation (i.e. ability to walk).

Will my data be kept confidential?

All information we receive from you will be treated confidentially. Creating a registry requires the existence of a file containing a patient's personal and medical data. This file will be subject to the regulations on data protection (national laws related to EU directive 95/46). The information will be encrypted and stored on a secure server.

If we publish any research or other documents based on the data from the registries, this research will never identify you by name. Third parties wishing to have access to the data in the registry (such as researchers or companies planning clinical trials or conducting research on new therapies) will only have access to anonymous information identifiable by a code. Before they are granted access even to this anonymous information, they have to have permission from the ethics or Steering committee. Your data will not be made available to employers, government organisations, insurance companies or educational institutions, nor to your spouse, other members of your family or your doctor.

A description of this study will be available on <https://www.clinicaltrialsregister.eu> and on <http://www.clinicaltrials.gov>. These websites will not include information that can identify you. At most, these websites will include a summary of the results. You can search these websites at any time. Your data will be kept for up to 15 years on a physically and logically secure cloud based server.

How will I benefit from registering?

The registry is intended as a public service for the benefit of patients living with HIBM. You will not receive any payment or any other financial benefit as a result of submitting your data to the registry. The results of research facilitated by the registry may be patentable or may have commercial potential. However, you will not receive patent rights and will not receive financial benefits from any future commercial development. Nevertheless, there may be other benefits from participating, including the following: we will inform you (on the basis of the information that you and your doctor provide) if you might be a suitable candidate for a certain clinical study or trial. We will also inform you if we receive any new information on your disorder which might be of interest to you, for example, if we find better ways for caring for patients with HIBM. The data collected might also provide benefits for other patients with your disorder, for example, by providing information on how many people worldwide have the same condition, or providing information for researchers interested in the best standards of care for your disorder. It will also help raise awareness of the disease and could encourage additional research and development.

Will I be informed of new information relating to the study?

Any new significant findings related to the study will be communicated to you by a member of the study team. We will also inform you if we publish any reports or papers based on the data obtained through the registry. In any publications we will never identify you by name.

Do I have to participate in the registry and can I withdraw if I change my mind?

Your continued participation in this project is completely voluntary. There are no consequences to withdrawing from this study and your refusal to be in the study or your withdrawal from the study will involve no penalty or loss of benefits to which you are entitled. You may stop being in the study at any time without affecting the availability of your future medical care.

The data protection act grants you the right to access your own data and to rectify them or withdraw them completely at any time. Should you wish to withdraw your data from the registry you will be free to do so without having to provide any explanation. If you wish to withdraw, you should get in touch with the staff in charge of the registry. Contact details are provided below.

Can you be removed from the registry?

The study curator or sponsor may end your participation in this registry without your permission at any time, if it is in your best interest or if the registry is stopped. You will be notified of this decision.

What are the potential risks of being in the registry?

The registry will be stored in a secure database online. Registering over the internet may pose some risk to data security; however, significant efforts are put in place to ensure this does not happen. You do not give up any of your legal rights by signing this form. There will be no experimental study drug given and no tests or procedures will be performed therefore there are no potential risks, or side effects of participating.

Who should I contact if I have any other questions?

If you would like any additional information or need to tell us about any change in your data, or if you wish to withdraw your data from the registry, please contact

Chief Investigator: Professor Hanns Lochmüller

Tel: 44 (0) 1912418602;

Post: Institute of Genetic Medicine

University of Newcastle, International Centre for Life

Newcastle Upon Tyne, NE1 3BZ, UK

Dr Oksana Pogoryelova

Tel: 44 (0) 191 2418640

email: HIBM@treat-nmd.eu

Post: Institute of Genetic Medicine

University of Newcastle, International Centre for Life

Newcastle Upon Tyne, NE1 3BZ, UK