

## **Understanding Your Registry Participation and Consent FAQs**

We (The Joshua Frase Foundation) are asking you to enter personal and other information into the International Family Registry for Centronuclear and Myotubular Myopathies (which we will call The Registry). It is important that we explain what is involved and what will be done with the information you provide. This section contains answers to questions about the information we want, why we want the information, how it might benefit you and our community.

Consent is needed to join the registry. You will be asked to read the Registry consent form. After you read the form, you will be asked a series of questions to confirm that you understand the project. Then you will be asked to sign electronically. Parents or legal guardians can sign on behalf of their child or on behalf of an adult who does not have the ability to consent for him/herself. Participation is optional.

If you have questions that are not answered in this document, please contact the registry coordinator at: [connect@joshuafrase.org](mailto:connect@joshuafrase.org).

### **What is a registry?**

A registry is a place to store detailed information about affected individuals with a specific disease or syndrome. The information does not include names or identifiers. Each participant's information is stored under a code number. In this case, The Registry is for individuals with CNM/XLMTM. Establishing The Registry addresses two critical needs. First, scientists studying CNM/XLMTM need accurate, firsthand information to understand the demographics of our community. Second, scientists who are ready to begin research studies will be able to access The Registry to identify people who may be eligible to participate. The research may be on bettering our understanding these diseases or conditions. We also hope that there may be research studies on new diagnostic measures or treatments. If an affected individual looks like a good match for a scientist's research, the scientist will contact the International Family Registry for Centronuclear and Myotubular Myopathies with the patient's code number. The curator of The Registry will then contact the CNM/XLMTM participant or representative. Until permission is given, scientists will not contact families directly.

### **What are the benefits of collecting affected individual's information in a registry?**

There is no direct medical benefit to the affected individual from being in the Registry. Although there is no guaranteed benefit to any individual participating in the registry, it greatly benefits our community. By collecting information on affected individuals in The Registry, scientists can:

- Study why individuals have different symptoms.
- Study how certain treatments work and don't work.
- Study how to help medical professionals improve how they treat affected individuals with CNM/XLMTM.
- Speed up research in CNM/XLMTM by collecting information that scientists can use.

- Let affected individuals (or their family) know when they may be eligible for clinical research studies or clinical trials.

### **Whose data are you collecting in The Registry?**

The Registry collects data from individuals who have a diagnosis of CNM/XLMTM, including those who have passed away. The diagnosis must be confirmed via genetic testing and/or a muscle biopsy.

### **Who can consent?**

Affected individuals over the age of 18 who understand the consent form (and thus do not have a legal custodian) are eligible to join The Registry on their own. Otherwise, the legal guardian, parent, or custodian of the patient must provide consent for the affected individual to join. When a minor affected individual becomes 18 (and if they are able), consent will be obtained directly from the affected individual for continued participation.

### **What are the steps to filling out The Registry with affected individual's information?**

If you are the affected individual or the representative for the affected individual, you should finish reading this document, then decide if you want to participate in The Registry. If so, you will need to read the Informed Consent form and complete the registry form provided, answering all questions to the best of your ability.

If more than one family member is affected with CNM/XLMTM, a new consent/understanding of consent form and registry questionnaire will need to be completed for each individual. The registry questionnaire should take no more than 20 minutes to complete.

### **Where will the data go?**

All of the information you provide will be maintained on a secure server managed by Qualtrix and The Joshua Frase Foundation, and any information that could identify you and your family members will not be shared without your consent. As this patient registry is designed to identify patients for clinical trials and/or research studies, your de-identified information may be shared with members of the research community.

### **Who will have access to the Registry records?**

The International Family Registry for Centronuclear and Myotubular Myopathies is the guardian of the information contained within The Registry. The goal of The Registry is to share medical and other information with scientists and other researchers, while still protecting your privacy. This is done by removing the name, address and other "identifying" information. We call this "de-identified" data (information from which all personal identification has been removed). The registry can share this de-identified data with the CNM/MTM community, approved scientists, researchers, clinicians and bio-tech companies.

The Registry has to follow rules to protect information about you. Federal and state laws also protect your privacy.

### **Will I be expected to provide The Registry with additional information in the future?**

Yes. The Registry is most valuable for scientific research when it is kept up-to-date. Therefore, you will be asked to update your profile and information once a year. We will send out notifications annually to remind you. Please contact the coordinator at [connect@joshuafrase.org](mailto:connect@joshuafrase.org) if there is a change in the affected individual's contact information.

**I want to be involved in a clinical trial. If I register, is this guaranteed?**

Although one of the main goals of The Registry is to make it easier for affected individuals to participate in research, there is no guarantee that that you or your family member will be eligible for a trial.

Please note that even if the coordinators of a clinical trial believe that you might be eligible for the trial, based on the data about you stored in The Registry, it is still possible you do not meet the trial inclusion criteria after all. Please also be aware that if we inform you about the existence of a trial, this does not imply that we endorse it. In order to participate in any trial, you will need to fill out a separate informed consent form directly with the researchers conducting the trial.

Please remember that joining the Registry does not obligate you to participate in any clinical trial or study.

**I don't want to be involved in a clinical trial. Should I still register?**

Absolutely. We hope that you will still be willing to register, even if you don't want to take part in a trial. Your information may still be useful to researchers who are trying to learn more about patients with CNM/XLMTM. Global conscience ushers in change, even without participating in clinical trials, your information will give us better understanding of our community as a whole.

**Can I withdraw if I change my mind?**

Your participation in this project is entirely voluntary. Should you change your mind and wish to withdraw your data from The Registry, you will be free to do so without having to provide any explanation. Simply contact The Registry Coordinator and all of your data will be removed from the database. De-identified data accessed prior to your request for removal cannot be retrieved from researchers that have already accessed it.

**If I have given data or information to doctors, researchers, clinics or hospitals in the past, is it OK to give my data to The Registry now?**

Yes. We will be taking precautions to make sure information used is not redundant with data that may have previously been collected. Our goal with this registry is to ensure that there is no overlap with any other existing registries.

**Will it cost me anything to be in this Registry?**

No. There are no costs to join the registry.

**Is the Registry still affiliated with the NIH/NCATS Global Rare Disease Registry (GRDR)?**

From 2016-2018 the Registry was a member of a consortium of rare disease registries through the NIH called the GRDR. Unfortunately, the funding for the GRDR project has now ended and

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therefore the Registry is no longer submitting new data to the GRDR and the formal affiliation with the NIH/GRDR is no longer active.

### **Who should I contact if I have any questions?**

If there are questions concerning the registry, your participation, or to report any problems that result from your participation in The Registry, please contact The Registry Coordinator at [connect@joshuafrase.org](mailto:connect@joshuafrase.org).