



## **NDF GNE Myopathy Patient Registry Frequently Asked Questions**

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

It is a collection of standardized information about individual persons who share a condition which is collected in a systematic and comprehensive way and can serve a variety of purposes. Shared informational data allows for a deeper and broader understanding of the nature of a disease and its patient population.

### **Why do you need a patient registry and how is it different from other registries?**

NDF needs a registry to help researchers and doctors learn more about GNE Myopathy and develop a better understanding of the disease (at what age it starts and how) and to know the numbers of the patient population globally. NDF is the largest nonprofit organization in the world working specifically on GNE Myopathy (HIBM). As such, it is vital that we continue in our efforts to build the most comprehensive database of patients ever created. Potential researchers, biotech companies and health care professionals require data in order to better understand the disease, develop new therapies and improve standard of care of GNE Myopathy patients. Our registry is an important component in our efforts to continue building our coalition of experts working on this disease.

### **Who is eligible?**

Anyone, from any region of the world who has been diagnosed with GNE Myopathy/GNEM (previously known as HIBM, Hereditary Inclusion Body Myopathy, Nonaka disease, Quadriceps Sparing Myopathy or SQSM, Distal Myopathy with Rimmed Vacuoles, Inclusion Body Myopathy or IBM2) is eligible to participate.

### **What information is collected and how is it stored?**

We will collect personal data such as name, age, sex, your mutation/s in the GNE gene, contact details, when you were first diagnosed, what symptoms you noticed, when they started and any clinical trial you participated in. We will also ask you to include details about your doctor to be able to direct other patients in your country to healthcare practitioners who are familiar with GNE Myopathy. Your information is collected in an electronic form and a unique code will be assigned to your data to guarantee it is anonymized.

### **Is there a cost to register and will I be compensated for participating in the registry?**

There is no cost to register and you will not receive any compensation for being part of the registry. You will not receive any royalties obtained from patents or commercial product developed as a result of the information that you provide in the registry. However, participation in the registry may help improve current standard of care for you and other patients and could result in the development of new therapies for the treatment of GNE Myopathy. Participation in the registry may also identify you as a potential candidate for future clinical trials.

### **Do I have to be 18 to register?**

No, you don't have to be 18 years of age or older to be part of the registry. However, if you decide that you want to be included in the registry and have not yet reached legal age, your parents or legal guardian needs to give their permission for you to register. Once you reach age 18, we will contact you to ask you whether or not you would like to continue participating in the registry.

## **Why should I participate in the NDF registry?**

Current advances in the scientific field have enabled the development of new potential therapies for the treatment of many diseases, including neuromuscular disorders. For some of those disorders, clinical trials are already on their way with promising results. Having a registry enables us to quickly identify the number of patients present worldwide, their specific mutation and their clinical stage. This information is particularly important for ultra-rare diseases such as GNE Myopathy, where only a few patients in the world will have the right profile for a therapeutic intervention such as a clinical trial. Having the patients' details, including each patient's particular genetic defect and other key information about their disease can be a powerful tool for physicians and health care professionals to understand current treatment practices, course of disease, factors that influence prognosis and quality of life.

## **Why is NDF collecting this information?**

- o Collecting information about as many GNE Myopathy patients as possible help scientists and health care professionals determine and predict the number of identified cases of GNE Myopathy worldwide in support of research and potential biotech industry partnerships.
- o Collecting information helps our scientific collaborators increase their understanding of the patient population and the disease, support design for potential natural history studies and clinical trials for new treatments, assist with the development of recommendations for standards of care and characterize the GNE Myopathy population as a whole.
- o Having patients' contact information allows NDF to be in touch with patients about important programs, studies and trials specifically tailored to their community.
- o Learning more about where GNE Myopathy patients live helps us identify regions where more attention is needed.

## **Is my data secured and who will have access to it?**

NDF is the guardian of the information contained in the Registry. We follow the US HIPAA and European GDPR guidelines for registrants' data protection. Only NDF staff and program managers who are part of the registry team have access to the secure data center that contains personal information. All information you provide will be encrypted and stored in a secure server. When requested and warranted for specific studies, NDF may share this information with scientific collaborators who request this data to move their studies forward. These requests are approved by NDF's Registry Advisory Committee, which is comprised of GNE Myopathy research experts. Patients' information is de-identified when shared with any qualifying NDF scientific collaborator. De-identified means that your name and contact information are removed from your data before being shared with others. Researchers and healthcare practitioners working on GNE Myopathy will only be granted access to de-identified information. NDF is committed to protecting your personal information.

Information included in the NDF registry will never be sold.

## **Can I remove my personal information from the registry if I change my mind and decide that I no longer want to participate?**

Participation in the registry is voluntary. You can contact NDF to withdraw your consent and remove your personal information at any time. If you decide to do so, your information will be permanently deleted from our database. You can re-enter the registry at any time. To withdraw from the registry contact: [registry@curehibm.org](mailto:registry@curehibm.org).

### **Can NDF remove me from the registry?**

NDF may decide to remove you from the registry for reasons that are not your fault. For example, if you were misdiagnosed and you don't have GNE Myopathy, your information would be removed. Removal of all your information from the registry will not affect your relationship with NDF. You can still be part of the NDF community. If your information is removed from the registry, you will be notified by email or mail.

### **I previously donated biological samples at one of your conferences to other Institutions, do I still need to register and will my data collected thus far be included in the registry?**

We ask all GNE Myopathy patients to register, regardless of past participation in other study protocols. This is a separate registry. Genomic, metabolomics and proteomic data obtained through biological specimens collected by other Institutions as part of standard of care or research studies are not part of the registry. Your data and medical records collected through other studies or by third parties will not be linked to this registry.

### **Does my doctor need to be involved in my participation?**

While we may ask who your doctor is in order to be able to share this information with other patients living in your area in need of a referral, there is no requirement to have a doctor involved in your participation.

### **Will I be informed of any results arising from the use of my data?**

Because the data shared with pharmaceutical companies or government institutions are de-identified, no one will know who the data belongs to and therefore, there is no way to share results on an individual basis. However, any new major finding obtained through the use of the database will be posted on the NDF website, thus providing real-time updates on the progress made in research.

### **Will NDF notify me when my data is shared with other parties?**

No, you will not be notified every time your de-identified data are shared with others. However, you can request to know if your data were shared with other parties and the list of who NDF shared it with.

Should you have any further questions about the NDF Patient Registry or your participation in it, please contact the Registry Coordinator:

By email: [Registry@cureHIBM.org](mailto:Registry@cureHIBM.org)

By mail: NDF 269 S. Beverly Drive, Suite 1206, Beverly Hills, CA 90212

By telephone: 310-721-1605

