NDF Informed Consent & GNE Myopathy Patient Registry

We appreciate your taking the time to complete NDF's GNE Myopathy Patient Registry Survey as we assemble a comprehensive patient database of patients with GNE Myopathy. Before you agree to take part in this registry, it is important that you read and understand why we are requesting this information and how the information will be used. Please review the FAQ document viewable and downloadable on our website that provides details about this registry. If after reading this, you have additional questions, please don't hesitate to contact the Registry Coordinator at: registry@curehibm.org.

We request that you fill out as much of the information as possible. We may contact you again over time, as we collect additional information.

By completing the survey, you give the Neuromuscular Disease Foundation consent to add your information to and contact you about the NDF GNE Myopathy Patient Registry with the understanding that patient information is being anonymized if shared with any qualifying NDF scientific collaborator. Your name and contact information will never be given to anyone without your permission.

Required Question(s)

Participation in this registry is voluntary. If you do not wish to be part of this registry, your relationship with NDF will not be affected.

By checking yes below, you consent/agree to the following:

I have read and I understand the information provided in the NDF GNE Myopathy Patient Registry FAQ sheet (available for viewing and download on our website curehibm.org/for-patients)

I have had the opportunity to ask questions, and all my questions have been answered to my satisfaction.

I consent to having my personal and clinical data stored in the NDF GNE Myopathy Patient Registry and to share my anonymized clinical data for research or clinical purposes and for the planning of clinical trials.

I agree to be contacted by NDF if additional information are needed and to receive follow-up forms in order to keep my record and the registry up to date.

I agree to inform NDF of any major changes in my personal or clinical data (for example change of address or changes in my medical condition, such as loss of ability to walk unassisted).

- ◯ Yes, I agree to all of the above.
- ◯ No, I do not agree to all of the above.

NEUROMUSCULAR DISEASE FOUNDATION
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Please enter the information indicated below.

By entering my personal information, I consent to receive email communications from NDF based on the information collected.

First Name: Last Name:

Home Phone: Email Address:

Address 1:

Address 2:

City:

State/Province (US/Canada):

Postal Code: Country:

Have you been diagnosed with GNE Myopathy?

- Yes
- No

Are you 18 years of age or older?

If you are under the age of 18 or responding on behalf of a minor, upon completion of this survey, please contact NDF at registry@curehibm.org for consent paperwork. Registration for minors cannot be accepted without this documentation.

- Yes, I am over the age of 18
- No, I am not over the age of 18

What is your date of birth? For example: May 10, 1978

Please select your gender

- Male
- Female
- Non-binary/third gender
- Prefer to self-describe in comments section
- Prefer not to say/Prefer not to answer: Comment:

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*** What is your ethnicity/nationality?**

*** When and at what age did you first notice symptoms?**

*** What were your first symptoms?**

*** Name and contact information of doctor who diagnosed you with GNE Myopathy.**

*** Are any of your siblings or relatives affected with GNE Myopathy?**

- ○ Yes, I have family members diagnosed with GNE Myopathy.
- ○ No, I do not have family members diagnosed with GNE Myopathy.
- ○ I prefer not to say.

*** Do you know the mutation(s) you have for GNE Myopathy? If so, please list your mutation(s) here. If you do not know your mutation, please write "I do not know".**

For example, 647T>C (V216A) in exon 4; 1892C>T (A631V) in exon 11.

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*** Please indicate which clinical trial(s) you have participated in:**

- [ ] NIH: A Natural History Study of Patients with GNE Myopathy
- [ ] NIH: Phase 1 Clinical Trial of ManNAc in Patients with GNE Myopathy or Hereditary Inclusion Body Myopathy (HIBM)
- [ ] NIH: A Phase 2 Study to Evaluate the Dose and Pharmacodynamic Efficacy of Sialic Acid-Extended Release (SA-ER) Tablets in Patients With GNE Myopathy or Hereditary Inclusion Body Myopathy (GNE Myopathy)
- [ ] NIH: An Open Label Phase 2 Extension Study of Higher Dose Sialic Acid-Extended Release (SA- ER) Tablets and Sialic Acid-Immediate Release (SA-IR) Capsules in Patients With Glucosamine (UDP-N-acetyl)-2-Epim-
  erase (GNE) Myopathy
  - [ ] NIH: An Open Label Phase 2 Study of ManNAc in Subjects With GNE Myopathy
  - [ ] NIH: Intravenous Immune Globulin to Treat Hereditary Inclusion Body Myopathy
- [ ] Ultragenyx: GNE-Myopathy Disease Monitoring Program (GNEM-DMP): A Registry and Prospective Observational Natural History Study to Assess GNE Myopathy or Hereditary Inclusion Body Myopathy (HIBM)
- [ ] Ultragenyx: Safety and Pharmacokinetics of Sialic Acid Tables in Patients With Hereditary Inclusion Body Myopathy (HIBM)
- [ ] Ultragenyx: Phase 3 Randomized, Double-Blind, Placebo-Controlled Study to Evaluate Sialic Acid in Patients With Glucosamine (UDP-N-acetyl)-2-epim-
  erase Myopathy (GNEM) or Hereditary Inclusion Body Myopathy (HIBM) (GNEM)
- [ ] Ultragenyx: Study to Evaluate the Safety and Efficacy of Aceneuramic Acid Extended-Release (Ace-ER) Tablets in Patients With Glucosamine (UDP-N-acetyl)-2-epim-
  erase Myopathy (GNEM) or Hereditary Inclusion Body Myopathy (HIBM)
- [ ] Ultragenyx: A Study to Evaluate the Safety of Aceneuramic Acid Extended Release (Ace-ER; UX001) Tablets in Glucosamine (UDP-N-acetyl)-2-Epim-
  erase (GNE) Myopathy (GNEM) (Hereditary Inclusion Body Myopathy) Patients With Severe Ambulatory Impairment
  - [ ] Tohoku University: Pharmacokinetic Study on N-acetylneuraminic Acid
  - [ ] Ludwig-Maximilians - University of Munich: Natural History in CCFDN and IBM Syndromes
- [ ] Institut de Myologie, France: Clinical, Biological and NMR Outcome Measures Study for Hereditary Inclusion Body Myopathy Due to Mutation of UDP-N-acetylglucosami-
  ne 2-epimerase/N-acetylm-
  annosamine Kinase Gene (GNE) (ClinBio-GNE)
- [ ] Sanford Research: Rare Disease Patient Registry & Natural History Study - Coordination of Rare Diseases at Sanford (CoRDS)
- [ ] None

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Would you like to receive our quarterly newsletter?

- ◯ Yes
- ◯ No

How did you find out about the NDF GNE Myopathy Patient Registry?

Please list specific names in the comments section below.

- ◯ NDF website and/or email
- ◯ NDF event (ie. Patient Day, Webinar, or HUDDLE)
- ◯ NDF Certified Patient Advocate
- ◯ A member of NDF
- ◯ Other

Comment:

Once complete, please submit this to NDF at registry@curehibm.org.

By submitting this survey, you confirm that all of the data entered is true and accurate and give the Neuromuscular Disease Foundation consent to add your information to and contact you about the NDF GNE Myopathy Patient Registry with the understanding that patient information is de-identified if shared with any qualifying NDF scientific collaborator.

Thank you for your participation in our GNE Myopathy Patient Registry Survey. We value and appreciate your input to our patient database. If you need further information or if we can be of assistance to you, please don't hesitate to contact us.