



Neuromuscular Disease Foundation Survey

*Required Question(s)



1. Please enter the information indicated below.

By entering my personal information, I consent to receive email communications from the survey author's organization based on the information collected.

First Name:

Last Name:

Home Phone:

Email Address:

emailaddress@xyz.com

Address 1:

Address 2:

City:

State/Province (US/Canada):

Postal Code:

Country:



* 2. Do you have GNE Myopathy?

Yes

No



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

50 characters left.



* 4. Please select your gender.

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

Comment:

500 characters left.

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

350 characters left.

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

350 characters left.

***** 7. What were your first symptoms?

350 characters left.

* 8. Who diagnosed you with GNE Myopathy?

350 characters left.

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

350 characters left.

* 10. Are any of your siblings or relatives affected with GNE Myopathy? If so, please list any siblings or relatives affected with GNE Myopathy. If you prefer not to answer, please write "prefer not to answer."

350 characters left.

* 11. Do you know the mutation(s) you have for GNE Myopathy? If so, please list your mutation(s) here.

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

350 characters left.

* 12. Please indicate which clinical trials 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-Epimerase (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-epimerase 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-epimerase 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-Epimerase (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-acetylglucosamine 2-epimerase/N-acetylmannosamine Kinase Gene (GNE) (ClinBio-GNE)
- Sanford Research: Rare Disease Patient Registry & Natural History Study - Coordination of Rare Diseases at Sanford (CoRDS)
- None

* 13. Would you like to receive our quarterly newsletter?

- Yes
- No

* 14. How did you find out about the Patient Information Survey?

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:

500 characters left.

Finish