NEUROMUSCULAR DISEASE FOUNDATION
JOIN THE GLOBAL EFFORT TO CURE NEUROMUSCULAR DISEASES

Patient Information Form

In our effort to collect information from and suggest resources to GNE Myopathy patients, we ask that you kindly provide us with any information that you are comfortable sharing with us. **By submitting this form, you give the Neuromuscular Disease Foundation consent to add your information and contact you about the NDF Patient Registry.**

Name:_____

Address: _____

_____

Email address: _____

Phone Number: _____

Do you have GNE Myopathy? Yes

Date of Birth (For example: May 10, 1978):_____

Gender: _____

Ethnicity/Nationality:_____

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

What were your first symptoms? _____

_____

Who diagnosed you with GNEM? _____

Name and contact information of doctor who diagnosed you:_____

_____

Are any of your siblings or relatives affected? No

If so, please list their names:_____

Do you know your mutations? If so, please list your mutation(s) here.

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

_____

Would you like to receive our quarterly newsletter? Yes

Please indicate which clinical trials you have participated in:

☐ **NIH:** A Natural History Study of Patients with GNE Myopathy (ClinicalTrials.gov Identifier: NCT01417533)

☐ **NIH:** Phase 1 Clinical Trial of ManNAc in Patients with GNE Myopathy or Hereditary Inclusion Body Myopathy (HIBM) (ClinicalTrials.gov Identifier: NCT01634750)

☐ **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) (ClinicalTrials.gov Identifier: NCT01517880)

☐ **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 (ClinicalTrials.gov Identifier: NCT01830972)

☐ **NIH:** An Open Label Phase 2 Study of ManNAc in Subjects With GNE Myopathy (ClinicalTrials.gov Identifier: NCT02346461)

☐ **NIH:** Intravenous Immune Globulin to Treat Hereditary Inclusion Body Myopathy (ClinicalTrials.gov Identifier: NCT00195637)

☐ **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) (ClinicalTrials.gov Identifier: NCT01784679)

☐ **Ultragenyx:** Safety and Pharmacokinetics of Sialic Acid Tables in Patients With Hereditary Inclusion Body Myopathy (HIBM) (ClinicalTrials.gov Identifier: NCT01359319)

☐ **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) (ClinicalTrials.gov Identifier: NCT02377921)

☐ **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) (ClinicalTrials.gov Identifier: NCT02736188)

☐ **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) (Also Known as Hereditary Inclusion Body Myopathy [HIBM]) Patients With Severe Ambulatory Impairment (ClinicalTrials.gov Identifier: NCT02731690)

☐ **Tohoku University:** Pharmacokinetic Study on N-acetylneuraminic Acid (ClinicalTrials.gov Identifier: NCT01236898)

☐ **Ludwig-Maximilians - University of Munich:** Natural History in CCFDN and IBM Syndromes (ClinicalTrials.gov Identifier: NCT01902940)

☐ **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) (ClinicalTrials.gov Identifier: NCT02196909)

☐ **Sanford Research:** Rare Disease Patient Registry & Natural History Study - Coordination of Rare Diseases at Sanford (CoRDS) (ClinicalTrials.gov Identifier: NCT01793168)