Who can sign up to the registry?
We welcome registrations to the GNE myopathy online patient registry from patients of any age, who are genetically confirmed or clinically diagnosed with GNE myopathy.

To register please visit the study website: www.gnem-dmp.com

What happens with the information provided by GNE Myopathy patients?
Anonymous data gathered will be accessible to the medical and research community, patients, families and patient organisations in the form of a scientific paper or report, upon approval from the Steering Committee and Ethics Committee. It is hoped that this information will provide insight into the disease, and help drive clinical trials and research that could lead to better treatment strategies.

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For more information about the GNEM-DMP, please visit:
www.treat-nmd.eu/gne/overview
www.ultragenyx.com/patients/gnem/
www.clinicaltrials.gov
ID number NCT01784679

To contact us:
HIBM@treat-nmd.eu
What is GNE Myopathy?

GNE Myopathy, also known as Hereditary Inclusion Body Myopathy (HIBM), Quadriceps-Sparing Myopathy (QSM), Distal Myopathy with Rimmed Vacuoles (DMRV), Nonaka Myopathy or IBM Type 2 is a rare, severe and slow progressive genetic muscle disease caused by mutations in the GNE gene.

GNE myopathy (GNEM) leads to weakness and wasting of muscles in legs and arms. First symptoms normally occur in young adults (usually in their twenties or thirties), but a later onset has also been observed in some patients. Initially, increased tripping and difficulty in climbing stairs is noticed, because of foot drop (dropping of the forefoot due to weakness). The condition gets worse over time, and may lead to weakness of the upper leg muscles with difficulties climbing stairs or getting up from sitting, and weakness of the hands and shoulder muscles. However, quadriceps muscles typically remain strong even in late stages. Severity and rate of progression are highly variable even within families, but GNE myopathy often leads to disability and loss of ambulation in later life. The heart, the respiratory muscles in the lower part of the body are generally the first to be affected.

Who is involved?

The GNEM-DMP is a partnership between Newcastle University (United Kingdom) and Ultradynex Pharmaceutical Inc. (USA) designed to improve the medical knowledge of GNE Myopathy. The Steering Committee includes GNE myopathy experts and patient organisation representatives to ensure that the partnership is always acting in the patients’ best interests. To view a list of Steering Committee members please visit: www.treat-nmd.eu/gne/patient-registries/steering-committee

Our research approach

To better understand GNE myopathy we have created a program, which combines online data collection via a registry and in-clinic data collection via a natural history study. This allows us to gather comprehensive information on the clinical presentation and progression of the disease. This information will be collected for several years.

Patient Organisations & Support

Below you will find a list of some of the patient organisations and support groups that cover GNE myopathy and Muscular Dystrophy:

- **Advancement of Research for Myopathies (ARM)**
  - www.arm.org

- **Muscular Dystrophy UK**
  - www.musculardystrophyuk.org

- **Neuromuscular Disease Foundation (NDF-USA)**
  - www.ndf-hibm.org

- **GNE Myopathy International**
  - www.gne-myopathy.org

- **Tara Talks GNE Myopathy**
  - www.taratalksgnemyopathy.blogspot.co.uk

- **GNE Myopathy In Focus**
  - www.gnemyopathy.com

Patient organisations and support groups that cover GNE myopathy to confidentially record and monitor their own health information (data) securely.

Who can participate?

There are no age restrictions on who can participate in the registry. However, you must have a diagnosis of GNE myopathy, be willing to provide your medical information and give electronic consent. If you are taking part in another registry, natural history study or clinical trial you can still participate.

What information will be collected?

The questionnaires you complete will ask you about your disease, general medical history, medications, quality of life, ability to move and muscle biopsy/genetic testing (if applicable). When you register, you will be assigned an online profile where you can can view all of the questionnaires you have completed during your participation.

The health information that you enter for the GNEM-DMP registry will be entered into an international database, which is supervised by TREAT-NMD. Your personal health information will be treated confidentially and will only be identified by an anonymous code, and not by your name. There are data protection measures in place to protect the information entered. The personal and medical information collected will be stored on a secure server for up to 15 years.

The study team will contact you after 6 months, 12 months and then yearly thereafter for up to 15 years, to ask you to update your information if you choose to do so.

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*These organisations are an incomplete listing of rare disease support organisations and are not controlled by, endorsed by, or affiliated with Ultradynex Pharmaceutical Inc. The list is meant for informational purposes only and is not intended to replace your healthcare professional’s medical advice. Ask your doctor or nurse any questions you may have about your disease or treatment plan.

The GNE Myopathy Online Patient Registry  www.gnem-dmp.com

The online patient registry is a way to follow how GNE myopathy affects (physically and emotionally) those diagnosed, monitor their health and and also aide researchers in designing clinical trials and potential treatments. The registry also allows people with GNE myopathy to confidentially record and monitor their own health information (data) securely.

Who can participate?

There are no age restrictions on who can participate in the registry. However, you must have a diagnosis of GNE myopathy, be willing to provide your medical information and give electronic consent. Even if you are taking part in another registry, natural history study or clinical trial you can still participate.

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Sign up at: www.gnem-dmp.com. For Further Information please contact the study curator at: HIBM@treat-nmd.eu