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.

Why should I sign up?

For You The Patient

Help you track your health

Access reports showing disease dynamics

Anonymously compare yourself to a deiidentified group of patients GNE myopathy

For Your Doctor

To help clinicians better manage and support a patients understanding of GNE myopathy

For Treatment Development

To help researchers design (and recruit for) clinical trials more quickly

To develop potential treatments

For The World

To help improve the standard of care for all people with GNE myopathy

To sign up to the GNE Myopathy Online Patient Registry visit:

www.gnem-dmp.com

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:

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gnemDMP
DISEASE MONITORING PROGRAM

Newcastle University · ultragenyx pharmaceutical · TREAT-NMD Neuromuscular Network

GNE Myopathy Disease Monitoring Program (GNEM-DMP)

A Registry and Prospective Observational Natural History Study to Assess GNE myopathy or Hereditary Inclusion myopathy (HIBM)

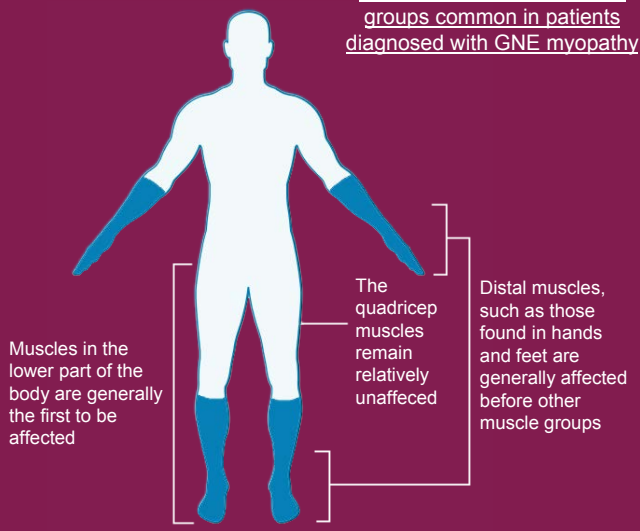
What is GNE Myopathy?

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 and speech and swallowing are usually not affected by GNE myopathy.

Overview of affected muscle groups common in patients diagnosed with GNE myopathy



Who is involved?

The GNEM-DMP is a partnership between Newcastle University (United Kingdom) and Ultragenyx 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 histrory 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 www.hibm.org

*Associazione Gli Equilibristi HIBM – (Italy) www.glieq uilibristi-hibm.org/

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

*Tara Talks GNE Myopathy - (USA) www.taratalksgnemyopathy.blogspot.co.uk/

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

*Muscular Dystrophy UK – (UK) www.musculardystrophyuk.org/

*Distal Muscular Dystrophy Patients Association - (Japan) www.npopadm.com

*Muscular Dystrophy Ireland (MDI) www.mdi.ie/

GNE Myopathy In Focus www.gnemyopathy.com

*These organisations are an incomplete listing of rare disease support organisations and are not controlled by, endorsed by, or affiliated with Ultragenyx 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.

gnemDMP DISEASE MONITORING PROGRAM

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.

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.

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