GNEM DMP Patient Fact Sheet

Title: Hereditary Inclusion Body Myopathy -Patient Monitoring Program (HIBM-PMP):
A Registry and Prospective Observational Natural History Study to Assess HIBM Disease
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The disease/condition: GNE myopathy (also known as hereditary Inclusion body myopathy) is a rare, severe and progressive genetic muscle disease due to mutations in the *GNE* gene. Other names for the same condition are Nonaka disease, Quadriceps-sparing myopathy or distal myopathy with rimmed vacuoles. First symptom of GNE myopathy is often foot drop. Progressive worsening leads to difficulties climbing stairs or getting up from sitting, and weakness of the hands and shoulder muscles. Although researchers put a lot of effort in finding a drug for this disease, currently there is no drug approved for treatment.

Who is involved?

The GNE myopathy Disease Monitoring Program is a public-private partnership between Newcastle University (United Kingdom) and Ultragenyx Pharmaceutical Inc. (USA) and is designed to improve the medical knowledge of GNE myopathy. A study Steering Committee consisting of HIBM experts and Patient advocacy groups will be closely involved in overseeing this research and will ensure that the project is always acting in the patients best interests.

Our research approach: To better understand the disease we have created a program, which combines a Patient Registry with a Natural History Study. This allows us to gather comprehensive information on the clinical presentation and progression of GNE Myopathy over several years.

The GNEM Patient Registry: The GNEM registry is designed to collect patient reported information about the GNE Myopathy condition through a web based platform. Information on changes in muscle function, mobility and quality of life over time will be reported by patients directly over a period of several years. The registry aims to connect patients and families with doctors and researchers by making sure that patients' details are all collected in a single database or "registry" in a safe and ethical way. The GNEM Patient Registry is available worldwide for all GNEM patients who are over 18 years old.

Natural History Study: The Natural History study is performed by selected centres in Europe, Middle East and North America. Professionally reported results of clinical evaluations by physicians will provide high quality validated data. This data will be valuable in the design of future clinical trials.

Who can register? We welcome registrations in the **HIBM Patient registry** from all patients over 18 years of age with genetically confirmed or clinically diagnosed HIBM (also known as GNE myopathy, Nonaka disease, DMRV or quadriceps sparing myopathy). To register please follow the link www.gnem-dmp.com.

What happens with the information provided by HIBM patients?

Anonymous data gathered through the registry will be accessible to the medical and research community, patients, families and patient organisations upon approval from the Steering Committee and Ethical Committee. It is our hope that this information will provide insight into the disease, and help drive clinical trials and research that could lead to better treatment strategies.

To register with HIBM Patient Registry please visit: www.gnem-dmp.com
For more information on the GNEM-DMP contact: HIBM@treat-nmd.eu
For more information about TREAT-NMD or Ultragenyx Pharmaceutical Inc. please visit:
http://www.treat-nmd.eu http://www.ultragenyx.com