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GNE Myopathy Patient Registry

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Background: The GNE Registry is an **international, patient reported, disease specific database** which provides the neuromuscular community with a **valuable, flexible and sustainable resource of disease-specific information**. The registry can be used to **support planning and recruitment for clinical trials**, and to capture real-world patient data (including data that may contribute to **post marketing surveillance** and **standards of care**). The GNE Registry is managed by the John Walton Muscular Dystrophy Research Centre (JWMDRC, Newcastle University, UK) and adheres to the TREAT-NMD Global Database Oversight Committee (TGDOC) Charter.
The GNE Myopathy Registry helps to facilitate translational research. The registry is **available in 7 languages**, collecting data since March 2014 with over 350 participants from over 30 countries worldwide (https://treat-nmd.org/what-is-a-patient-registry/).

Aims:
- To contribute to the disease understanding by collecting reliable information
- Facilitating clinical trial readiness and translational research by helping the recruitment of patients to clinical trials
- Helping to overcome the scarcity of resources and the geographic isolation of patients
- Informing patients and the GNE myopathy community of latest developments in scientific research and disease management, via newsletters.

Methods: Patient reported data gathered through disease-specific questionnaires (GNEM-FAS, quality of life (SF12) and other non-validated questionnaires).

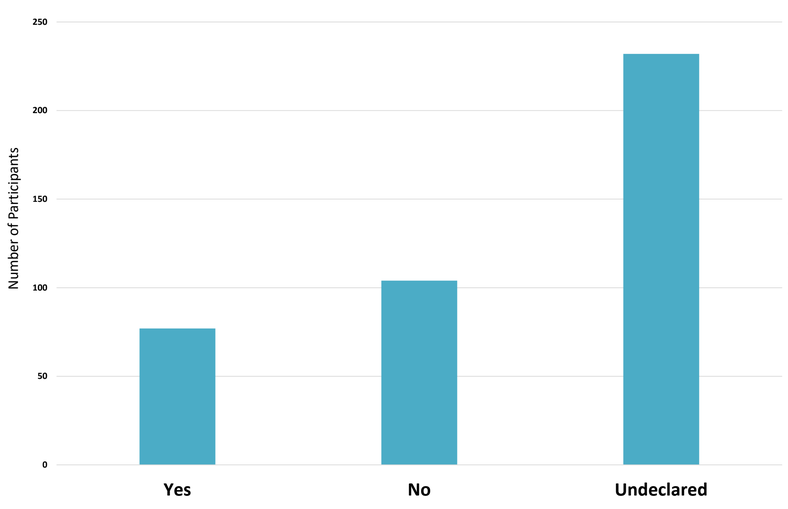


Figure 1 shows the number of registry participants who require walking support

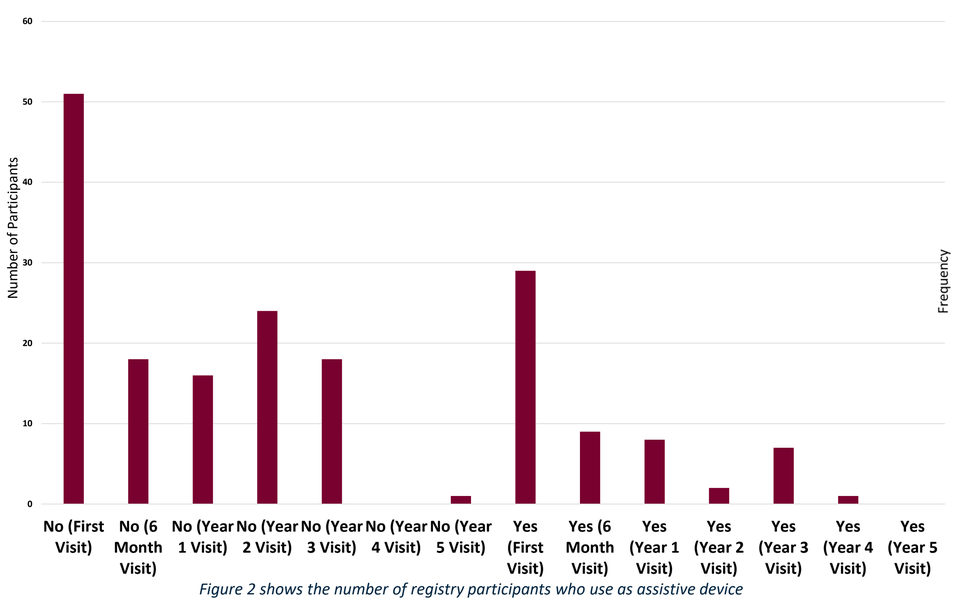


Figure 2 shows the number of registry participants who use as assistive device

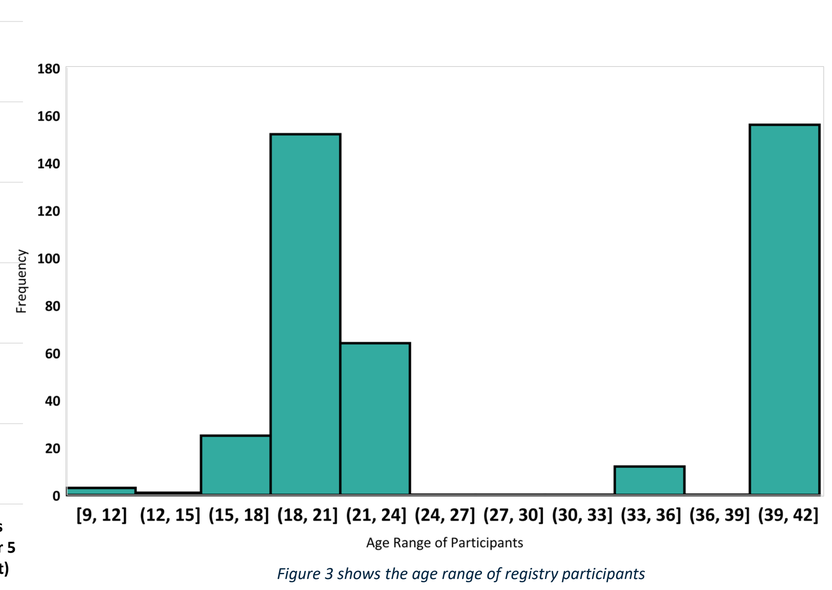


Figure 3 shows the age range of registry participants

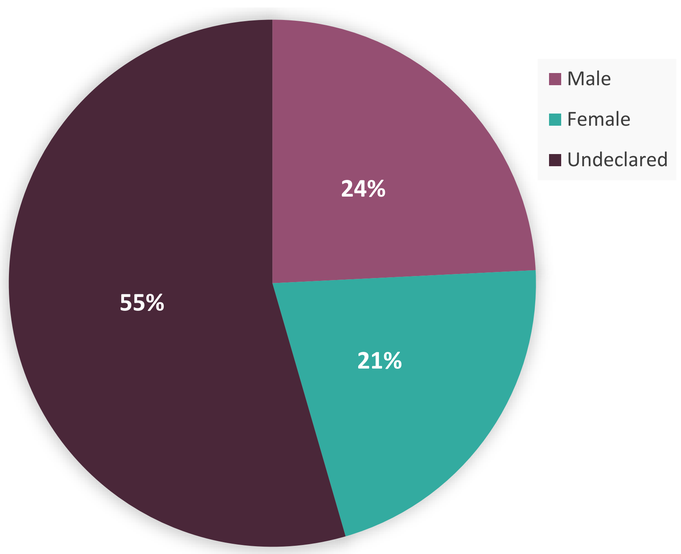


Figure 4 shows the sex of registry participants

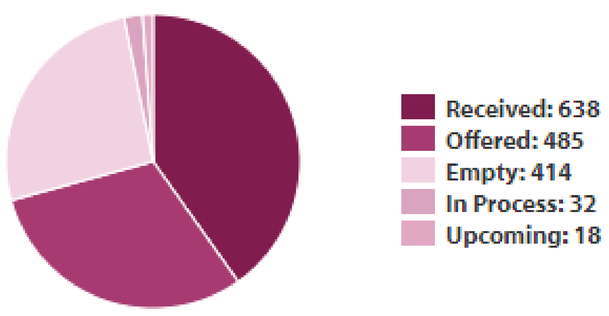
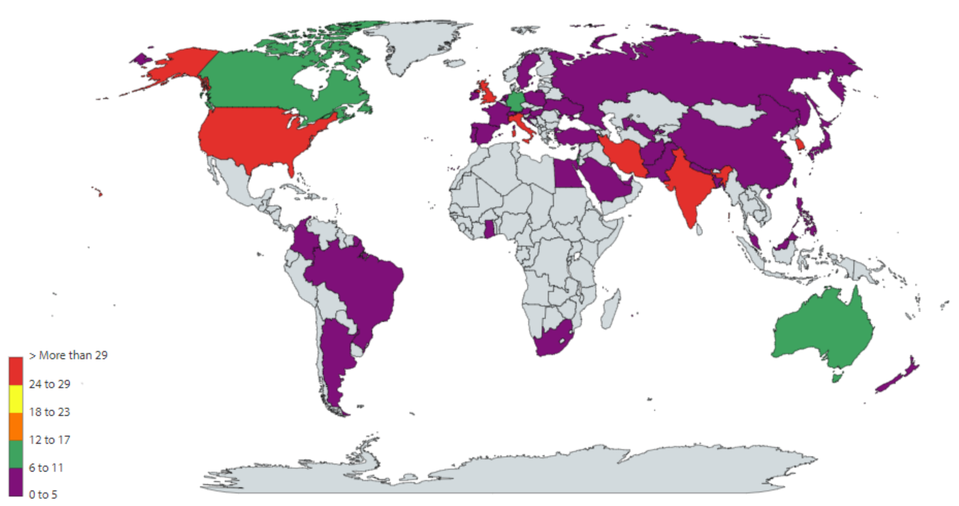


Figure 5 shows the questionnaire compliance of registry participants

The map below shows the participation by country for the GNE Registry. The legend below explains the color gradations.



The total number of participants in the GNE Registry is 393

Results: The registry has enabled a genotype-phenotype study to be conducted. Previously, this platform played an important role in:
a. Contributing to the disease understanding by collecting reliable information;
b. Facilitating clinical trial readiness and translational research by supporting the recruitment of patients into clinical trials;
c. Helping to overcome the scarcity of resources and the geographic isolation of patients;
d. Informing patients and the GNE myopathy community of latest developments in scientific research and disease management, via newsletters and the TREAT-NMD Network.

The registry helps to facilitate recruitment for patient advocacy meetings, clinical trials (NCT02736188, NCT02377921) and natural history study (NCT01784679). The registry actively participates in EURORDIS and MDUK research, namely The Voice of Rare Disease Patients and GNE Patient's Day.

Conclusions: The International GNE Myopathy Patient Registry is a valuable tool for this ultra-rare disease community

For more information, contact the registry curator at lucy.imber@ncl.ac.uk or hibm@treat-nmd.org

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