Introduction:

The TREAT-NMD Global Alliance is an independent network governed by an Executive Committee of academic, clinicians and patient representatives who facilitate collaborative research in neuromuscular disease (NMD). This international not-for-profit network is also governed by a Charter which outlines membership requirements for those who wish to join the Alliance.

The network aims to accelerate drug development, provide new therapies to patients swiftly and improve access to relevant information on standards of diagnosis and care.

One of the key TREAT-NMD infrastructures is the Global Registry Network, governed by the TREAT-NMD Data Systems Oversight Committee. The network is a federation of individual, independent, national or regional patient registries where members collect agreed disease specific datasets. The FSHD Global Registry Network collects data from 21 registries, representing four continents (Figure 1).

Method:

An electronic survey requesting demographic and diagnostic data was sent to all TREAT-NMD member registries, collecting FSHD data in 2022.

Results:

There were 13 (824) survey responses from registries in Australia, Belgium, Czech Republic, Denmark, Germany, Japan, Latvia, Netherlands, New Zealand, Slovenia, Turkey (2 registries) and UK.

Collectively the registries provided data on 3,572 FSHD patients: 1,528 were female, 1,645 male and gender was not reported in 199 cases. Only 90 patients (3%) were aged <16 years.

Most patients had FSHD1 (1,471/1,563; 55%) with fewer FSHD2 (82, 3.5%) cases. However, 42% of patients (1,344) were unknown FSHD type. Overall, 43% of patients (1,463) received genetic confirmation of FSHD. With FSHD1 cases (1,262/1,747; 72%) expected higher than FSHD2 (32/82, 39%) or unknown FSHD type (17/1,334, 1%).

Figure 2

Responses from 13 registries provided data on 3,572 patients. Registries provided data on: (a) FS HD subtype; (b) number of patients aged 16 and under vs over 16; and (c) whether a diagnosis was genetically confirmed for each subtype of FSHD.

Conclusions:

The TREAT-NMD FSHD Global Registry Network represents an international harmonised data resource, providing opportunities for researchers and industry to support clinical trial planning upon its interrogation. Despite most registries being clinician reported (62%), there were many patients without FSHD genetic confirmation or a specific FSHD type diagnosis. Understanding these aspects nationally will be important as they represent clinical trial essential criteria.

If you are interested in understanding more about the TREAT-NMD Global Registry Network and how interrogating this data could support your research or drug development efforts, please contact registries@treat-nmd.com.