The UK FSHD Patient Registry: Data Collection and Collaboration

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Background

- The UK FSHD Patient Registry is a patient self-enrolling online database established in May 2013 to collect clinical and genetic information about FSHD type 1 (FSHD1) and FSHD type 2 (FSHD2)
- Supported by Muscular Dystrophy UK (MDUK) and coordinated by the John Walton Muscular Dystrophy Research Centre at Newcastle University
- Core Member of the TREAT-NMD Global Registry Network
- The registry's primary aim is to facilitate and accelerate clinical research in FSHD
- The registry also aims to better characterise and understand FSHD, share information on standards of care, and disseminate information relating to upcoming academic, clinical and non-clinical studies

Method

- The registry is used to capture **longitudinal**, **self-reported** data through an online portal available to patients and specialist clinicians.
- **Patient reported outcomes** are entered into a secure portal, combined with clinician verified genetic details. Patients are reminded to update their information annually, and all registrations are confirmed by the registry curator.
- Data collected within the registry includes all mandatory and highly encouraged items in the TREAT-NMD Core Dataset for FSHD v2, including both **patient reported**, and **doctor reported items** such as genetic confirmation.
- The registry is also now able to receive **genetic reports** directly from patients via a secure file upload link.
- The registry also captures the **McGill Pain Questionnaire**, an **FSHD Pain Questionnaire**, **the Short Form 36 Health Survey Questionnaire (SF-36)**, the **Individualized Neuromuscular Quality of Life Questionnaire**, and a **Scapular Fixation Questionnaire**.
- The registry can support researchers and industry with a wide range of projects, including creation of **de-identified patient data reports** for use in feasibility studies, dissemination of research surveys, and trial recruitment support.

Demographics

Patient Numbers

As of Nov 2022, there are **887** active UK based participants enrolled on the UK FSHD Patient Registry.

This includes 448 male and 439 female participants (a 51/49% split).

The average age is 51.8 years (Male = 52.3 years, Female = 51.1 years).

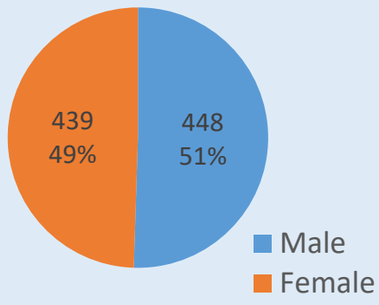

Figure 1. Self-reported patient sex at registration

Clinical / genetic diagnosis

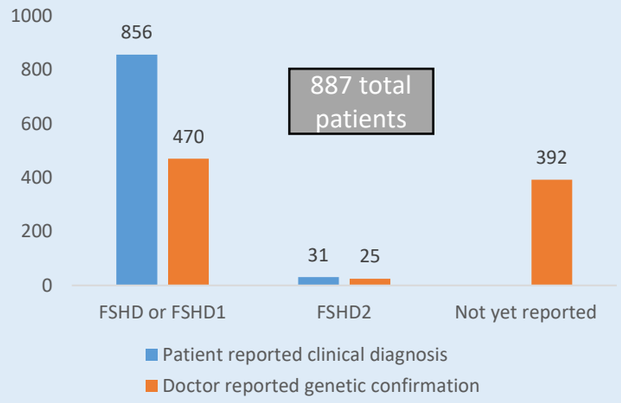

Figure 2. Self-reported clinical diagnosis and doctor reported genetic confirmation of diagnosis

The most common self-reported **clinical diagnosis** is FSHD/FSHD1 (96.5%) followed by FSHD2 (3.5%). It is expected this will move closer to the 95%-5% split reported in literature once more genetic confirmations are received.

Genetic confirmation has so far been provided for **56%** of all patients, and extensive communication and registry development is underway to increase this.

Family History

62% percent of patients reported a **positive family history** of FSHD, with 31% reporting no family history, and 7% unknown.

Patients' mothers were reported to be affected in 27% of the cohort, compared to 17% of their fathers.

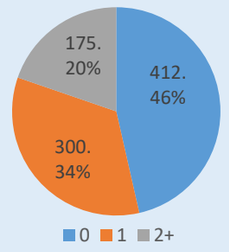

Figure 3. Number of affected immediate family members

Of patients' **immediate families**, 46% of patients reported no affected family members, 34% reported at least one affected family member, and 20% reported at least two affected family members.

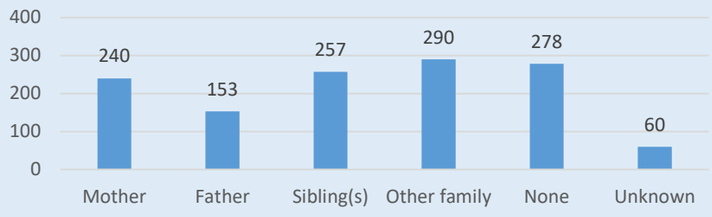

Figure 4. Family members affected with FSHD

Longitudinal data and completeness

501 registered patients **(57%)** have updated their registry record over the last 24 months, and a further 189 **(21%)** have updated in at least the last 5 years.

Longitudinal data is available for at least 455 **(51%)** of patients to date.

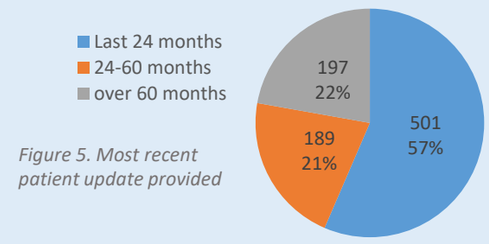

Figure 5. Most recent patient update provided

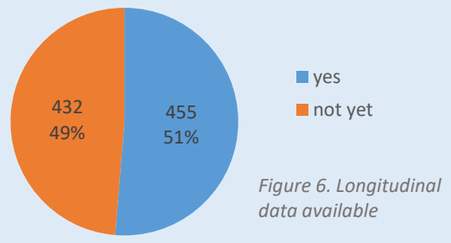

Figure 6. Longitudinal data available

Registry Use and Engagement with TREAT-NMD

To date the UK FSHD Patient Registry has supported at least **32** enquiries from industry, academics, clinicians and patient organisations. Most registry enquiries have involved online survey distribution (63%) supporting data analysis (16%), or research study recruitment (13%) **(Figure 13).** Since 2020, the registry has supported 14 surveys. For transparency and to highlight the versatility of the registry, enquiries that the registry has supported are now documented on the registry website.

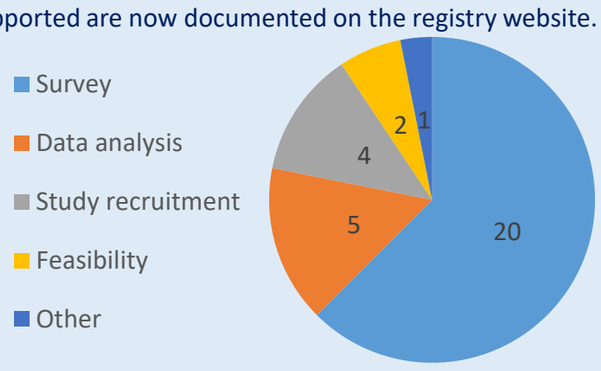

Figure 13. The type and number of registry enquiries supported since 2013

Future Engagement and Collaborations

Ensuring the registry dataset is fit for purpose is vitally important to ensure appropriate and relevant data is available for research. The UK FSHD Patient Registry is currently undergoing a dataset revision process in order to align with other TREAT-NMD registries, implement FAIR data principles, and other international standards and agreed data elements. The registry will also be keen to support efforts by TREAT-NMD to further update their Core Dataset for FSHD.

Core Dataset

The UK FSHD Patient Registry collects all the mandatory and highly encouraged patient-entered items in the TREAT-NMD Core Dataset for FSHD v2, based on the agreements made at the 171st ENMC International Workshop. The registry is also a Core Member of TGDOC; this allows the registry to participate in Global Registry Enquiries coordinated by TREAT-NMD.

Global Registry Enquiries

The UK registry was delighted to participate in the first TREAT-NMD Global Registry Enquiry involving FSHD in 2022, providing aggregate data to an industry partner for the purposes of feasibility and clinical trial design. Results will be shared on the registry website when available.

Other TREAT-NMD collaborations

The UK registry was also pleased to be included in TREAT-NMD's first poster describing the FSHD Global Registry Network, presented at the FSHD International Research Congress 2022. Of the 3,372 FSHD patients reported in the global network, approximately 26% are located within the UK registry.

The TREAT-NMD Core Dataset for FSHD

The TREAT-NMD Core Dataset for FSHD (v2) includes data elements on symptom onset, current best motor function, wheelchair use, ventilation support, retinal disease, hearing loss, pregnancy and scapular fixation. The charts below indicate a current snapshot of this data in the UK registry:

The majority of patients (91%) reported **weakness** in the shoulder (male – 94%, female – 90%), with considerably more females than males reporting facial weakness (76% to 61%) Only 3% of all patients reported no symptoms (male – 2%, female – 5%).

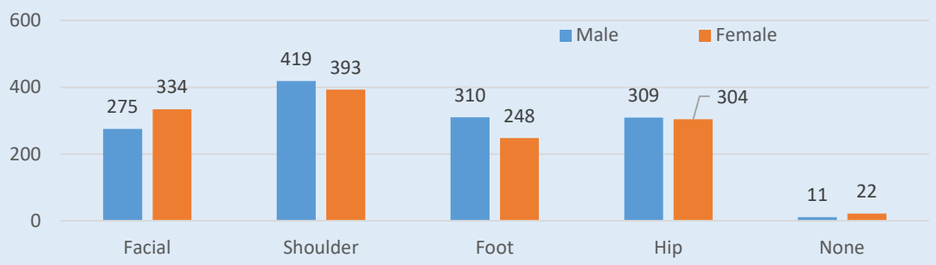

Figure 14. Number of patients reporting muscle weakness by sex

The average age of patient self-reported **symptom onset** ranges from 22 years (facial weakness), to 35 years (hip weakness). The most commonly described symptom present from birth was facial weakness, reported by 50 patients (0.06%). The maximum age for symptom onset reported was shoulder weakness at 79 years of age.

Symptoms reported	Facial	Shoulder	Foot	Hip
At birth	50	13	8	7
Average Age	22	26	33	35
Max Age	73	79	74	74

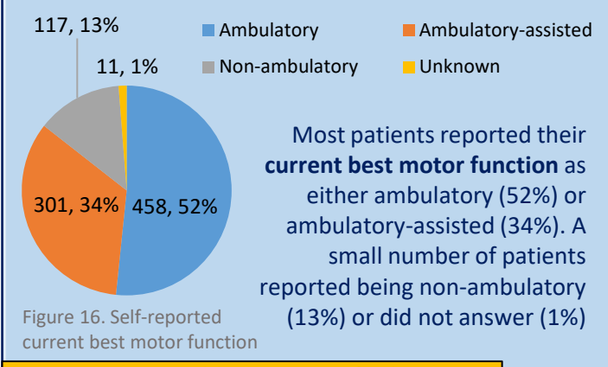

Figure 16. Self-reported current best motor function

Most patients reported their **current best motor function** as either ambulatory (52%) or ambulatory-assisted (34%). A small number of patients reported being non-ambulatory (13%) or did not answer (1%)

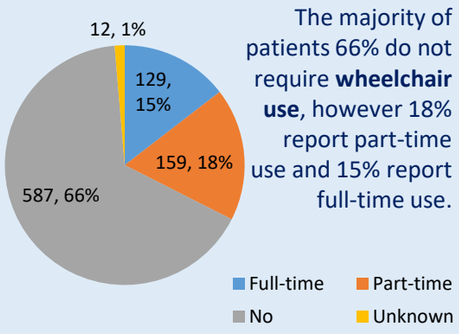

Figure 17. Self-reported wheelchair use

The majority of patients 66% do not require **wheelchair use**, however 18% report part-time use and 15% report full-time use.

Non-invasive ventilation was reported by 6.3% of patients (full time – 0.4%, part-time – 5.9%), whilst 1.3% of patients report using invasive ventilation (full time – 0.2%, part-time – 1.1%). The vast majority of patients reported no invasive (93%) or non-invasive ventilation (89%).

Retinal vascular disease was reported by only 1% of patients, with 84% reporting no retinal issues.

Hearing loss was reported by 16% of patients with 69% reporting no hearing issues

Scapular fixation surgery was reported by 8% of patients (bilateral – 5%, unilateral – 3%). 87% of patients reported they have had no scapular fixation surgery

Contact the UK Registry

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Registry Website
https://bit.ly/ukfshdreg

Email the curator
fshdregistry@newcastle.ac.uk

Use the registry data in your research
https://bit.ly/fshddata

Acknowledgement to Muscular Dystrophy UK for their continued support and to all the patients and clinicians who continue to participate in the registry.

The UK FSHD Patient Registry continues to be a versatile, cost-effective research tool that has helped facilitate a range of studies and advance FSHD research around the world. Additional work continues to be done to update the registry questionnaires, improve engagement with more doctors in the UK and increase the reporting of genetic information on the registry. As well as supporting research projects, the registry continues to develop new and engaging communication materials for the FSHD community and plans to further capture the patient voice in the development of new materials.