Clinical Features of the UK FSHD Patient Registry

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Thanks to our registry funders:

Muscular Dystrophy UK – Fighting muscle-wasting conditions

UK FSHD Patient Registry

Email the registry curator

Background

The UK Facioscapulohumeral Muscular Dystrophy (FSHD) Patient Registry is a patient self-enrolling online database collecting clinical and genetic information about FSHD type 1 (FSHD1) and type 2 (FSHD2). The registry was established in May 2013 with support from Muscular Dystrophy UK and is coordinated by Newcastle University.

Aims

The registry aims to facilitate academic and clinical research, better characterise and understand FSHD, and disseminate information relating to upcoming studies and research advancements.

Method

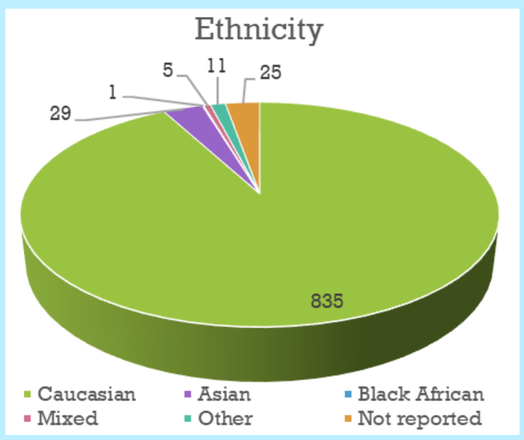
The registry captures longitudinal, self-reported data through an online portal available to patients and clinicians. Where specialised clinical or genetic information is required, the neuromuscular specialist involved in the patient's care can be invited to provide some additional information and the patient can select them from a pre-populated list at the registration stage. The registry is a Core Member of the TREAT-NMD Global Registries Network for FSHD.

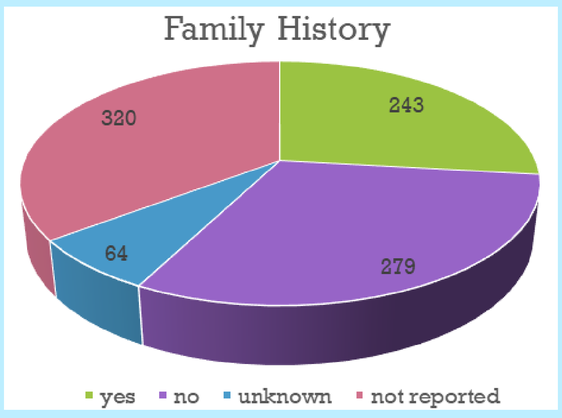
Results: As of March 2023, there were 906 active, UK based patient registrations. Data is also available for an additional 252 patients who are inactive or not based in the UK (their data is not included here). For those reporting a clinical diagnosis, 95.4% have FSHD or FSHD1, and 3.3% have FSHD2. Overall, 58.1% of patients have had genetic confirmation of their condition. In addition to collecting specific genetic data inputted by clinicians, the registry is now able to receive digital copies of patient's genetic reports directly via a secure upload portal. The registry has supported 33 registry enquiries to date, recent examples including a large Health Economics project, a survey on UK service provision, and various surveys capturing information on patient preferences, dysphagia, pregnancy, sleep, and the patient/caregiver experience.

Demographics

The ages of registry participants range from 6 to 88 years, with an average age of 51.8 years. 50.1% of patients are male and 49.9% are female.

The majority of registry participants reported their ethnicity as Caucasian (92.2%). Other ethnicities reported as Asian (3.2%), 'other' (1.2%), Mixed (0.6%) and Black African (0.1%).

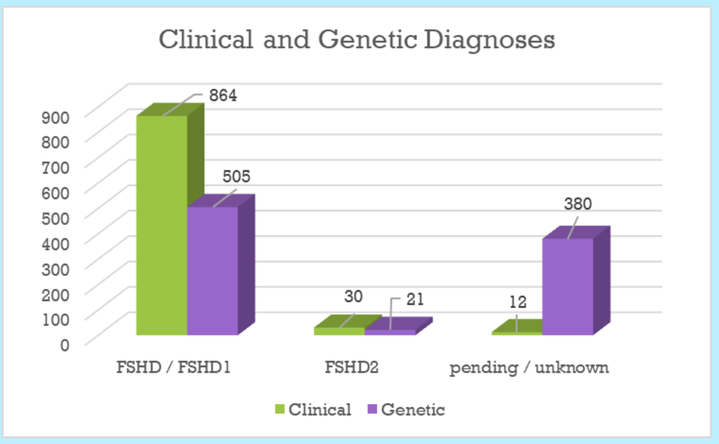


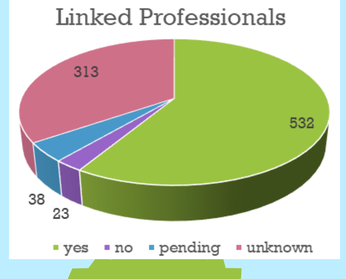


26.8% of patients reported a history of FSHD in at least one family member, whereas 30.8% reported no known family history. Positive family history was reported in 26.5% of patients' mothers, 17.9% of fathers, 28.8% of siblings, and 32.1% in another family member

Diagnoses

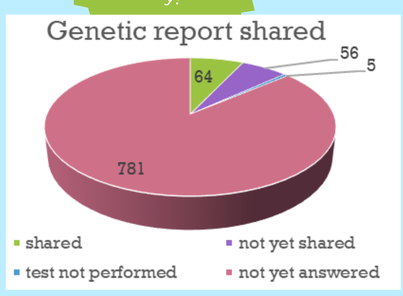
The most common patient-reported clinical diagnosis is FSHD or FSHD1 affecting 95.4%, and 3.3% reporting FSHD2. 1.3% are unsure or awaiting confirmation of their diagnosis. Genetic confirmation of diagnosis is now available for 58.1% of all registry participants.





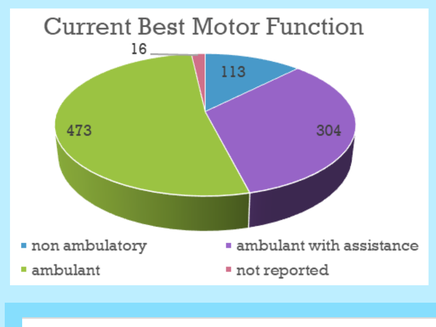
9.4% increase in linked professional users since March 2022!

Genetic report upload is a **new feature** of the registry!

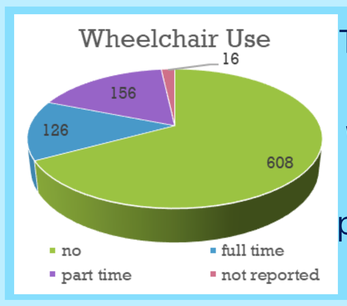


58% of registry participants now have a linked professional user (neuromuscular consultant, genetic counsellor, physio etc.) to verify patient-entered data and confirm genetic reporting. 2.5% do not see a specialist, and 4.2% have a professional user with a pending invitation. Full genetic test report has been shared by 7.1% to date.

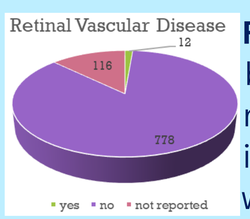
Clinical features



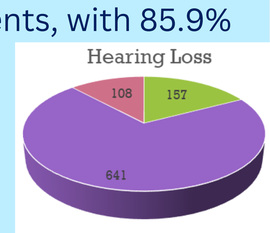
Most patients reported their current best motor function as either ambulatory (52.2%) or ambulatory-assisted (33.6%). A small number of patients reported being non-ambulatory (12.5%).

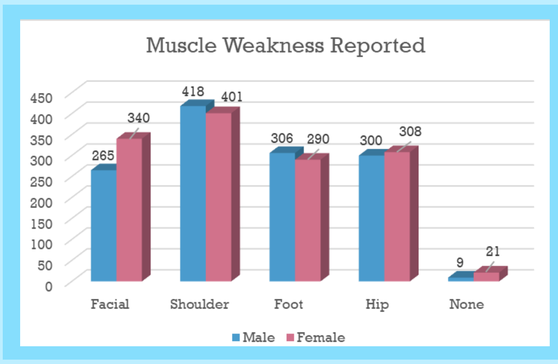


The majority of patients do not require wheelchair use (67.1%), however 17.2% report part-time use and 13.9% report full-time use.

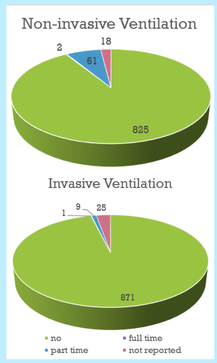


Retinal vascular disease was reported by only 1.3% of patients, with 85.9% reporting no retinal issues. **Hearing loss** was reported by 17.3% of patients with 70.8% reporting no hearing issues



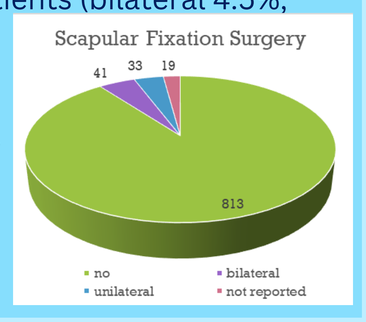


The majority of patients (90.4%) reported weakness in the shoulder (male 92.1%, female 88.7%), with considerably more females than males reporting facial weakness (75.2% to 58.4%) Only 3.3% of all patients reported no muscle weakness (male – 2%, female – 4.6%)



Non-invasive ventilation was reported by **7%** of patients (full time 0.2%, part-time 6.7%). Only **1.1%** of patients report using **invasive ventilation** (full time 0.1%, part-time 1%). The vast majority of patients reported no invasive (96.1%) or non-invasive ventilation (91.1%).

Scapular fixation surgery was reported by 8.2% of patients (bilateral 4.5%, unilateral 3.6%). 89.7% of patients reported they have had no scapular fixation surgery



Conclusion

The UK registry is currently one of the largest national FSHD patient registries globally and is an example of a versatile, cost-effective research tool, helping facilitate and advance a wide range of FSHD research. The new genetic report upload feature is shown to be improving the genetic information available on the registry, alongside the increase in neuromuscular specialist signing up as professional users. There are plans to review and update the patient questionnaires in the near future, and data linkage plans between the registry and the Newcastle Research Biobank for Rare and Neuromuscular Diseases which will enable more data to be available to facility research into FSHD. Additional work around patient engagement and promotion of the registry to neuromuscular specialists is required to increase the number of patients signing up to the registry, and efforts are required to increase the diversity of the registry population.