The UK FSHD Patient Registry: a powerful tool to support clinical research and patient voice in the translational research pathway

Helen Walker[1], Richard Orrell[2], Andrew Graham[3], Fiona Norwood[4], Mark Roberts[5], Tracey Willis[6], Emma Matthews[7], Mark Mencias[7], Robert Muni-Lofra[1], Chiara Marini-Bettolo[1]

1. The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, UK
2. UCL Queen Square Institute of Neurology, University College London, London, UK
3. Patient Representative, UK
4. Department of Neurology, Kings College Hospital, London
5. Department of Neurology, Salford Royal NHS Foundation Trust, Salford
6. Neuromuscular Service, The Robert Jones and Agnes Hunt Orthopaedic Hospital NHS Foundation Trust, Shropshire
7. The Atkinson Morley Regional Neurosciences Centre, St George's University Hospital NHS Foundation Trust, London

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Muscular Dystrophy UK – Fighting muscle-wasting conditions

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.

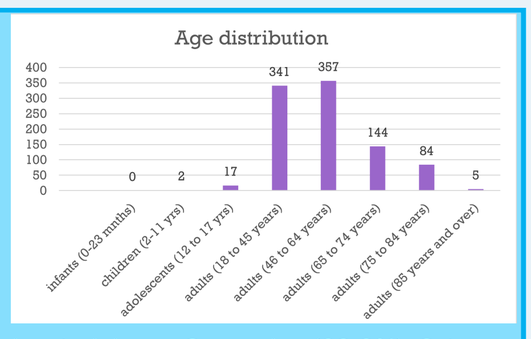
Method

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.

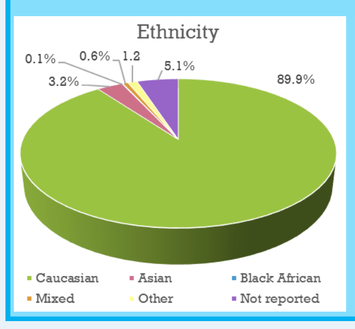
Results: As of May 2023, there were 950 active, UK based patient registrations. Data is also available for an additional 218 patients who are deceased, unresponsive or not based in the UK (their data is not included here). For those reporting a clinical diagnosis, 95.6% have FSHD or FSHD1, and 3.4% have FSHD2. Genetic confirmation has been reported by 56.8% of patients. 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 34 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.

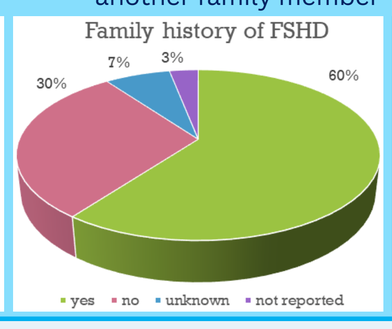
Demographics

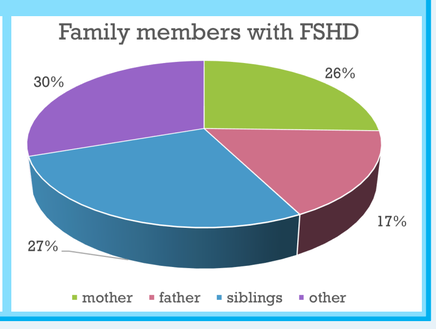
The ages of registry participants range from 6 to 88 years, with an average age of 51.4 years. Adults (age 18-64) comprise 73.5% of the participants, with elderly (age 65+) making up 24.5%, and paediatric (under 18) totalling 2% of participants. Sex is evenly distributed; 50.1% of patients are male and 49.9% are female.



The majority of registry participants reported their ethnicity as Caucasian (89.9%). Other ethnicities reported as Asian (3.2%), 'other' (1.2%), Mixed (0.6%) and Black African (0.1%). 5.1% did not report their ethnicity. A history of FSHD in at least one family member was reported by 60% of patients , whereas 30% reported no known family history. Positive family history was reported in 26% of patients' mothers, 17% of fathers, 27% of siblings, and 30% in another family member

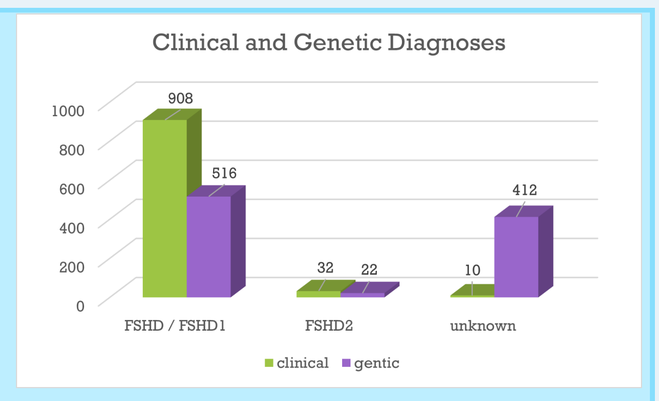


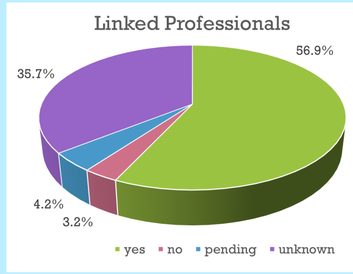




Diagnoses

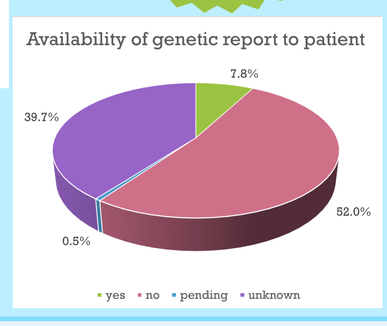
The most common patient-reported clinical diagnosis is FSHD or FSHD1 affecting 95.6% of participants, and 2.4% report FSHD2, and 1.1% are awaiting their diagnosis. Genetic confirmation of diagnosis has been recieved for 56.8% of all registry participants.





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

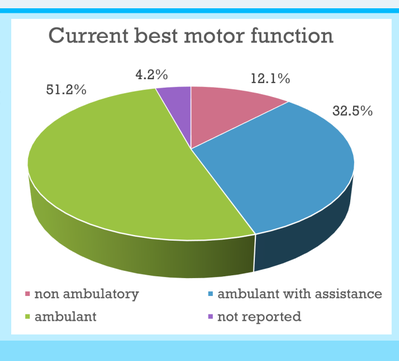
16% increase in genetic data reported since May 2022!

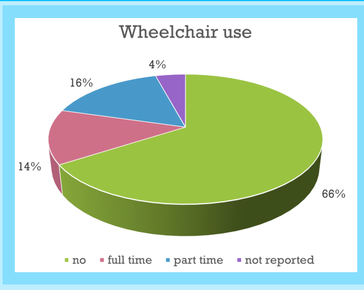


57% of registry participants now have a linked professional user (neuromuscular consultant, genetic counsellor, physio etc.) to verify patient-entered data and confirm genetic reporting. 3.2% do not currently see a specialist, and 4.2% have a professional user with a pending invitation. A genetic test report has been shared by 7.8% of patients to date, with 52% reporting they do not currently have access to their report.

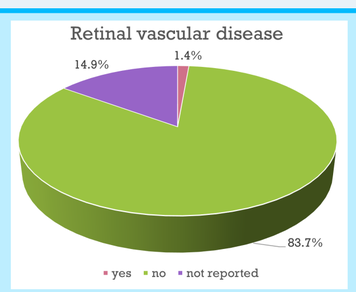
Clinical features

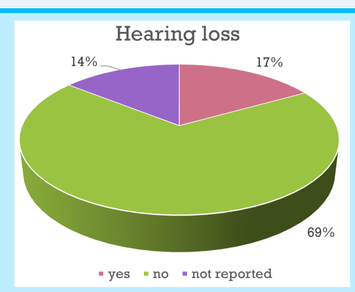
Most patients reported their current best motor function as either ambulatory (51.2%) or ambulatory-assisted (32.5%). A small number of patients reported being non-ambulatory (12.1%), and motor function was not reported by 4.2%



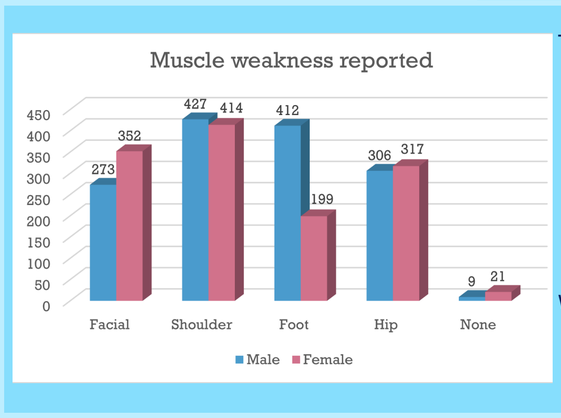


The majority of registry participants do not require wheelchair use (65.9%), however 16.3% report part-time use and 13.6% report full-time use. This data is not yet available for 4.2% of participants.



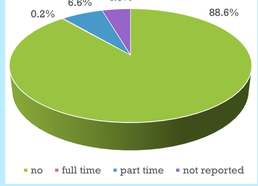


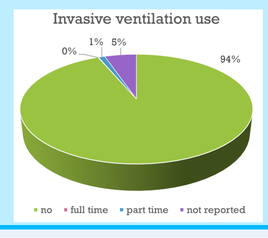
Retinal vascular disease was reported by only 1.4% of patients, with 83.7% reporting no issues. Hearing loss was reported by 16.8% of patients, with 68.8% reporting no hearing issues.



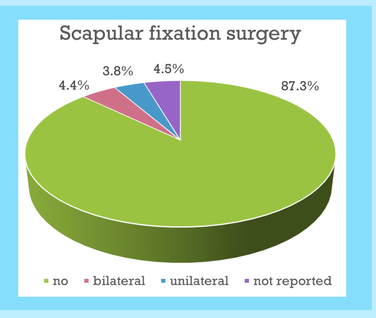
The majority of patients (88.5%) reported weakness in the shoulder (male 89.7%, female 87.3%). The biggest difference in symptoms reported by sex were in foot and facial weakness; considerably more males report foot weakness (86.6% / 42%), and more females report facial weakness (74.3% / 57.4%). Only 3.2% of all patients reported no muscle weakness (male – 1.9%, female – 4.4%)

Non-invasive ventilation was reported by 6.8% of patients (full time 0.2%, part-time 6.6%). Only 1.2% of patients report using invasive ventilation (full time 0.1%, part-time 1.1%). The vast majority of patients reported no invasive (93.6%) or non-invasive ventilation (88.6%).





Scapular fixation surgery was reported by 8.2% of all patients (bilateral 4.4%, unilateral 3.8%). 87.3% of patients reported they have had no scapular fixation surgery.



Conclusion

The UK registry is one of the largest national FSHD patient registries globally and is an example of a versatile, cost-effective research tool, helping to 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 specialists 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 are ongoing to increase the number of patients aware of and signing up to the registry, and efforts are required to increase the diversity of the registry population.

Meet the Registry Team

Ms Helen Walker
Registry Curator & Project Manager

Dr Chiara Marini Bettolo
Registry Principle Investigator

Email the registry curator

Registry Website
https://bit.ly/ukfshdreg

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