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

Helen Walker[1], Robert Muni-Lofra[1], Richard Orrell[2], Andrew Graham[3], Fiona Norwood[4], Mark Roberts[5], Tracey Willis[6], Emma Matthews[7], Mark Mencias[7], Kate Adcock[8], Chiara Marıni-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
8. Muscular Dystrophy UK, London, UK

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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. The registry also collects real-world evidence and supports data enquiries from industry and academia.

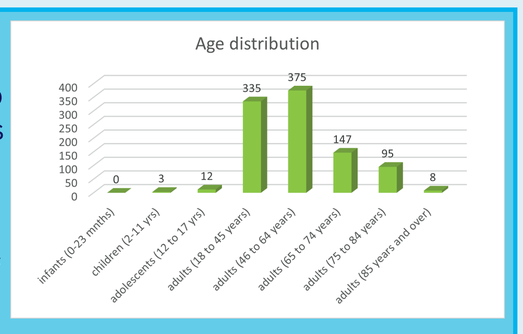
Method

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.

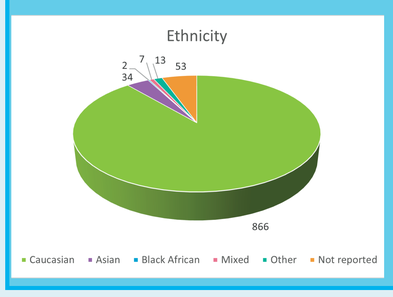
Results: As of April 2024, there were 975 active, UK based patient registrations. Data is also available for an additional 225 patients who are deceased, unresponsive or not based in the UK (their data is not included here). For those reporting a clinical diagnosis, 92.6% have FSHD or FSHD1, and 3.6% have FSHD2. Genetic confirmation has been received for 56% 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, to be entered by the registry curator. 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 - see registry website for details.

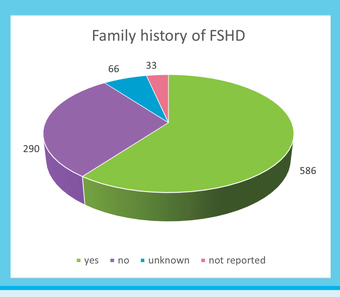
Demographics

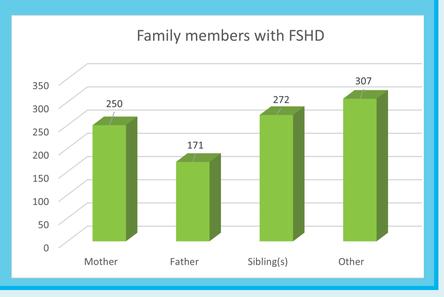
The ages of registry participants range from 5 to 89 years, with an average age of 52 years. Adults (age 18-64) comprise 72.8% of the participants, with elderly (age 65+) making up 25.6%, and paediatric (under 18) totalling 1.5% of participants. Sex is evenly distributed; 50.4% of patients are male and 49.6% are female.



The majority of registry participants reported their ethnicity as Caucasian (88.8%). Other ethnicities reported were Asian (3.5%), 'other' (1.3%), Mixed (0.7%) and Black African (0.2%). 5.4% did not report their ethnicity. A history of FSHD in at least one family member was reported by 60.1% of patients , whereas 29.7% reported no known family history. Positive family history was reported for 25.6% of patients' mothers, 17.5% of fathers, 27.9% of siblings, and 31.5% in another family member

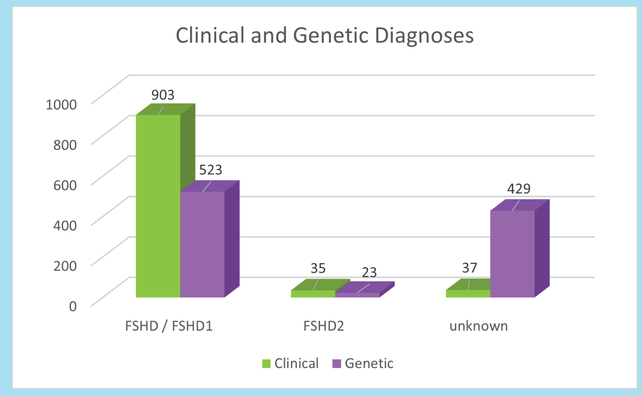


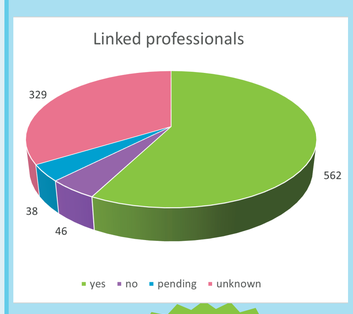




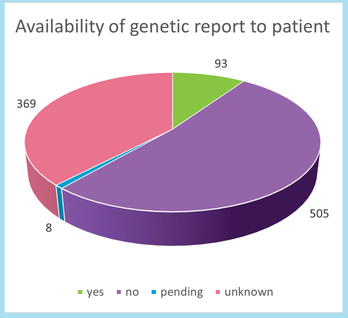
Diagnoses

The most common patient-reported clinical diagnosis is FSHD or FSHD1 affecting 92.6% of participants. A further 3.6% report FSHD2, and 3.8% have no yet reported their diagnosis. Genetic confirmation of diagnosis has been received for 56% of all registry participants.





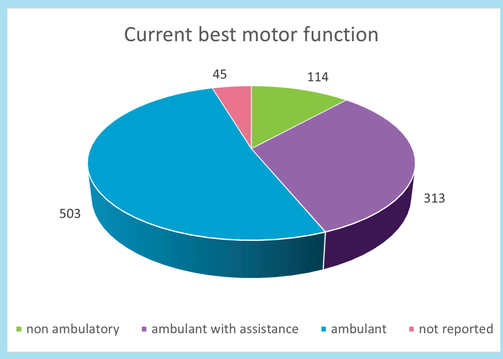
Now collecting genetic test report data!

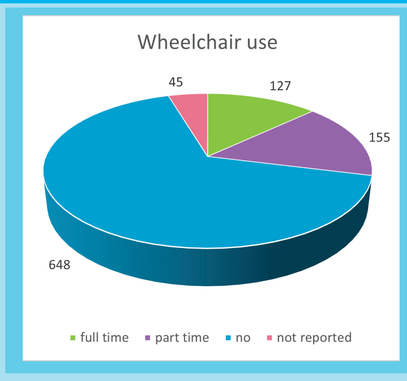


57.6% of registry participants now have a linked professional user (neuromuscular consultant, genetic counsellor, physio etc.) to verify patient-entered data and confirm genetic reporting. 4.7% do not currently see a specialist, and 3.9% have a professional user with a pending invitation. A genetic test report has been shared by 9.5% of patients to date, with 51.8% reporting they do not yet have access to their report.

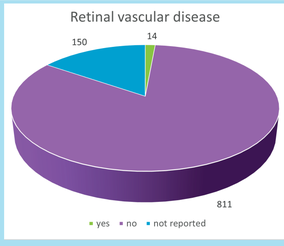
Clinical features

Most patients reported their current best motor function as either ambulatory (51.6%) or ambulatory-assisted (32.1%). A small number of patients reported being non-ambulatory (11.7%), and motor function was not reported by 4.6%.

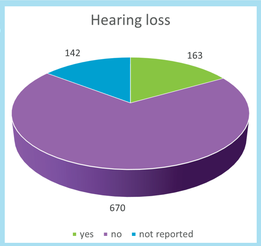




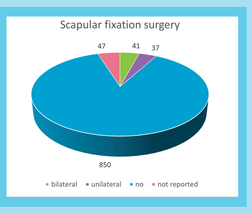
The majority of registry participants do not require wheelchair use (66.5%), however 15.9% report part-time use and 13% report full-time use. This data is not yet available for 4.6% of participants.

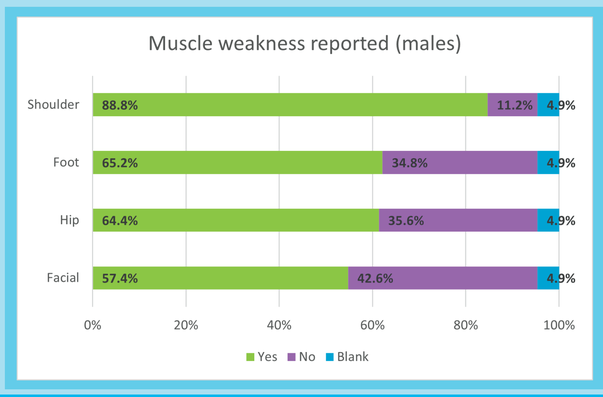


Retinal vascular disease was reported by only 1.4% of patients, with 83.2% reporting no issues. Hearing loss was reported by 16.7% of patients, with 68.7% reporting no hearing issues.

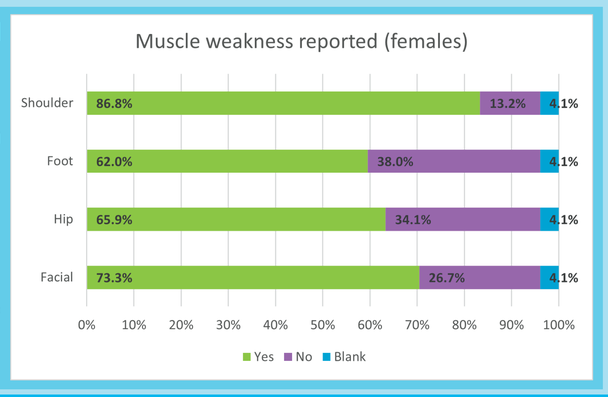


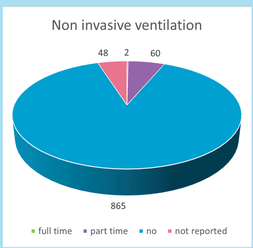
Scapular fixation surgery was reported by only 8% of all patients (bilateral 4.2%, unilateral 3.8%). Most participants (87.3%) reported they have had no scapular fixation surgery, and 4.8% did not answer.



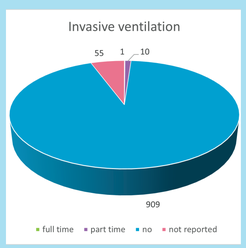


The majority of patients (87.8%) reported weakness in the shoulder (male 88.8%, female 86.8%). The biggest difference in symptoms reported by sex were found in facial weakness; considerably more females report facial weakness (73.3% / 57.4%), making this the second most highly reported symptom in women, as compared to foot weakness in men. 4.5% of participants did not answer the question.





Non-invasive ventilation was reported by 6.4% of patients (full time 0.2%, part-time 6.2%). Only 1.1% of patients report using invasive ventilation (full time 0.1%, part-time 1%). The vast majority of patients reported no invasive (93.2%) or non-invasive ventilation (88.7%).



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.

Contact Us

Ms Helen Walker
Registry Curator & Project Manager

Dr Chiara Marini Bettolo
Registry Principle Investigator

Registry Website
https://bit.ly/ukfshdreg

Email the registry curator