P599

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

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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 the John Walton Muscular Dystrophy Research Centre (JWMDRC) at Newcastle University.

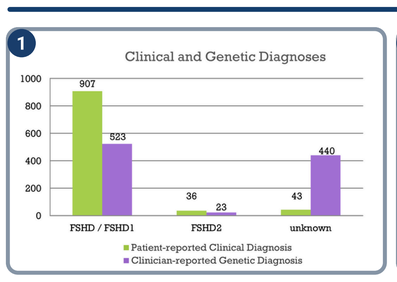
Aims

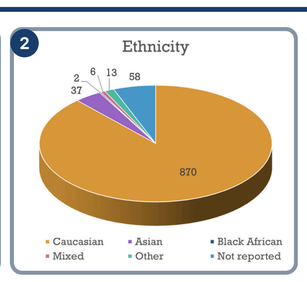
The registry facilitates academic and clinical research, enables better characterisation and understanding of FSHD, and disseminates information relating to upcoming studies and research advancements to participants.

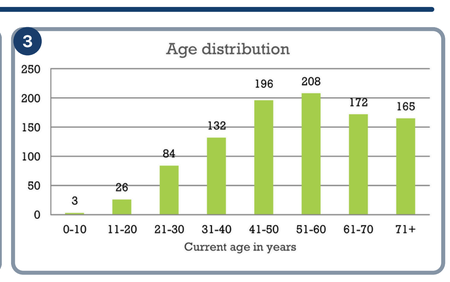
Method

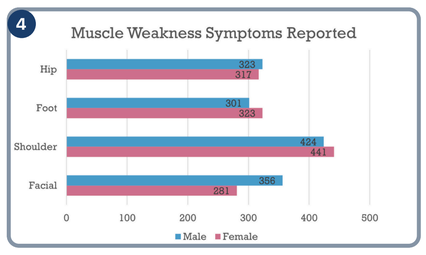
The registry captures longitudinal, self-reported data via a secure online portal. Where specialised clinical or genetic information is required, neuromuscular specialists involved in patients' care are invited to provide additional information. The registry is a Core Member of the TREAT-NMD Global Registries Network for FSHD, collecting the standardised core dataset and contributing to global data enquiries.

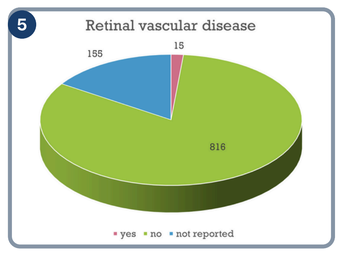
Figures

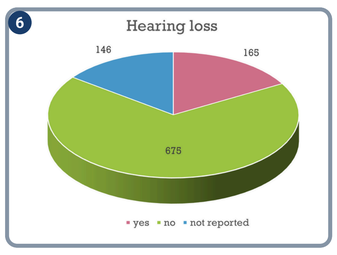


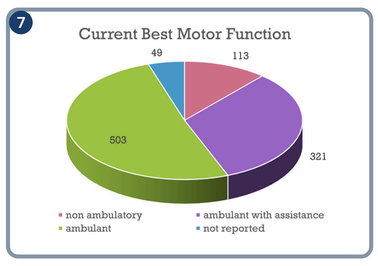


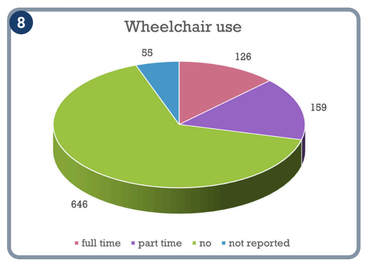


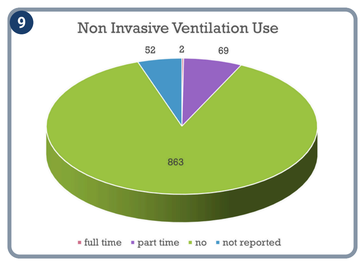












Results

As of September 2024, there are 986 active patient registrations in the registry. Data from an additional 228 deceased, unresponsive or inactive patients is not included in this analysis. For those reporting a clinical diagnosis, 92% have FSHD or FSHD1, 3.7% have FSHD2, and for 4.4% their diagnosis is pending or unknown [*fig.1*]. Genetic confirmation of the condition has been received for 55.4% of active patients to date. The majority of participants report their ethnicity as Caucasian (88%), 3.8% as mixed, and almost 6% did not answer [*fig.2*]. The average age of participants is 52 years, with a range between 5 to 90 years [*fig.3*], and sex is fairly evenly represented (49.7% female, 50.3% male). Shoulder weakness is the most highly reported symptom by 88% of participants; with hip (65%), facial (65% and foot (63%) weakness reported at similarly lower levels. Interestingly, there is a significant gender difference found in the data on facial weakness, with 16% more females than males reporting this symptom [*fig.4*]. Retinal vascular disease is reported by only 1.5% of patients *[fig.5],* and hearing loss reported by 17% *[fig.6*]. Most patients reported their current best motor function as ambulant (51%) or ambulant with assistance (33%) [*fig.7*]; and 29% reported using a wheelchair (16% part-time, 13% full-time) [*fig.8*]. Part-time non-invasive ventilation (NIV) was reported by 7% of patients *[fig.9]*, with only 0.2% requiring full-time NIV and 0.1% reporting invasive ventilation support. Please contact the registry manager to request more detailed data reports via the registry enquiry process.

Conclusions

One of the largest national FSHD patient registries and an example of a versatile, cost-effective research tool helping to facilitate and advance a wide range of research, the registry has supported around 40 registry enquiries. Additional work continues to be done to improve reporting of genetic information on the registry, and a dataset expansion project is currently underway to ensure data collected is relevant and useful. New data to be collected will include Patient-Reported Outcome Measures (PROMs), trial readiness, and access to care questions, and Privacy Protecting Record Linkage (PPRL) items to support anonymous data sharing. Ethnicity and other demographic questions will be refined in the updated dataset to allow for more accurate and detailed cohort information, and to inform PPIE activities to increase diversity, increase awareness of the registry in under-represented groups, and remove any barriers to participation. The registry is also transitioning to a new bespoke software platform incorporating automatic data validation and logic, data dashboards, analysis tools, and an improved user experience for patients and clinicians. These developments will support further patient recruitment and ensure registry data available to support research into FSHD is of the highest quality.

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MUSCULAR DYSTROPHY UK | OUR MUSCLES MATTER

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Meet The Team

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Registry Manager

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Principle Investigator

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