Advancing Neuromuscular Research: Achievements and Future Directions for the JWMDRC Patient Registries

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Background

There are unique challenges that come with conducting research into rare neuromuscular conditions, By collecting data on a national or international level from affected individuals, registries can help facilitate research in the neuromuscular field in several ways. Here, we demonstrate how the registries at the John Walton Muscular Dystrophy Research Centre have supported research since their inception. We also highlight developments planned for the registries to continually improve their value to the research community.

Participants

Registry	Participants (10/04/2025)	Genetically Confirmed	Male / Female	Median age (Range)
UK FSHD Patient Registry	1012	539	506 / 507	53 (8 – 88)
UK SMA Patient Registry	674	454	348 / 326	28 (0.5 – 85)
UK Myotonic Dystrophy Patient Registry	903	364	427 / 476	48 (0.5 – 87)
Global Registry for COL6-related dystrophies	379	159	187 / 192	19 (0.5 – 85)
Global FKRP Registry	1100	473	482 / 618	39 (1 – 85)
The Myotubular and Centronuclear Myopathy Patient Registry	557	301	353 / 204	23 (0.5 – 88)

Registry Process

Patient-initiated Registration
Registration is led by the patient, or parent/guardian for children. An appropriate eConsent form is signed before data entry.

Patient Selects Clinician
Patients can select their clinician and consent to them verifying their diagnosis and providing further clinical data.

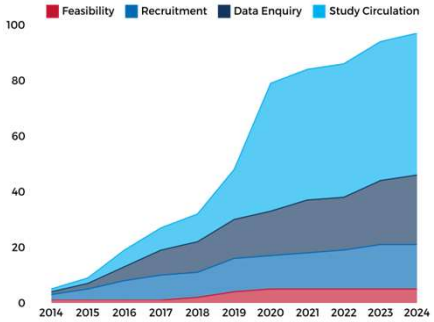
Patient and Clinician Data Entry
The Patient and Clinician complete the assigned questionnaires. These collect data including demographics, genetic diagnosis, and QoL.

Registry Enquiries reviewed by Steering Committee
Each registry Steering Committee is comprised of affected patients, patient advocacy representatives, and expert clinicians and researchers.

De-identified data supports research and trial recruitment
The registries can circulate information about clinical trials, support feasibiliy assessments, or provide de-identified data to researchers.

Registry Activity

Over the past 10 years, the JWMDRC Registries have completed 97 registry enquiries. These range from providing de-identified data to researchers, to providing targeted recruitment support services for trials and studies.



Case Study – UK SMA Patient Registry

The UK SMA Patient Registry has played a role in many stages of the translational pathway that has led to patients in the UK gaining access to both Nusinersen (Spinraza) and Risdiplam (Evrysdi). Most recently, the UK SMA Registry has supported the Managed Access Agreements (MAAs) for adult and paediatric SMA patients receiving Nusinersen or Risdiplam by collecting key PROMs data required by NICE.

Supporting Research Projects

Research projects can benefit from the data collection infrastructure established by existing registries, which can be adapted to include stand-alone research studies. This reduces duplication of data, resource and effort across the community. In collaboration with the MTM & CNM Patient Registry, the Liver Collaborative Working Group designed a liver function questionnaire which was added into the main registry questionnaire in 2023. This project was initiated by patient leaders in response to the urgent community need for more data on the involvement of the liver in these conditions. Publication of the group's finding is anticipated in 2025.

Future Directions

Work is underway to upgrade the patient registries to a new bespoke software platform developed by Newcastle University. Benefits will include improved features and functionalities, the ability to create new registries and sub studies, improved data quality and data completeness, and long-term sustainability savings. This upgrade has been planned alongside revisions and expansions to the UK FSHD and UK Myotonic Dystrophy Registry Datasets.

Initial platform development has been kindly funded by donations from a range of industry partners working in the neuromuscular space. This collaborative effort demonstrates the value of collaboration across the commercial field. We are grateful to our current donors:

Acknowledgements

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