Patient Registries and their Importance in Translational and Clinical Research

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Neuromuscular diseases (NMD) are a heterogeneous group of genetic conditions characterized by progressive muscle degeneration and weakness. In Europe, all NMD have a prevalence of ≤5 per 10,000 and are therefore considered rare. Patient registries are research databases containing demographic, genetic and clinical information about individuals affected by a condition or genetic mutation, and can facilitate translational research in rare neuromuscular diseases by:

- Rapidly locating participants for clinical trials and research studies.
- Providing natural history and epidemiological data.
- Answering specific research questions from academics, healthcare professionals and industry.
- Collecting real world data to inform regulatory pathways and monitor the safety and efficacy of new treatments.
- Informing the understanding and development of standards of care.
- Linking the research community to the patients by offering a two-way flow of diseasespecific information.

Patient registry data can be made available for academic or commercially funded research—please get in touch with the registries team to discuss how we can support your project.

Since 2008 the JWMDRC patient registries have been involved in over 50 scientific publications, and over 75 research enquiries including, but not limited to:

- Clinical trial recruitment
- Dissemination of clinical and academic research surveys
- Provision of de-identified data to researchers for analysis and publication.

Examples of registry enquiries include:

- ♦ Recruitment onto phase I, II and III clinical trials in multiple disease areas
- Obtaining patient feedback on a clinical trial protocol
- Recruitment onto non-interventional research studies on topics such as wearable activity monitors, accessibility issues in the built environment, and pregnancy in NMD.

Contact the JWMDRC registries team:



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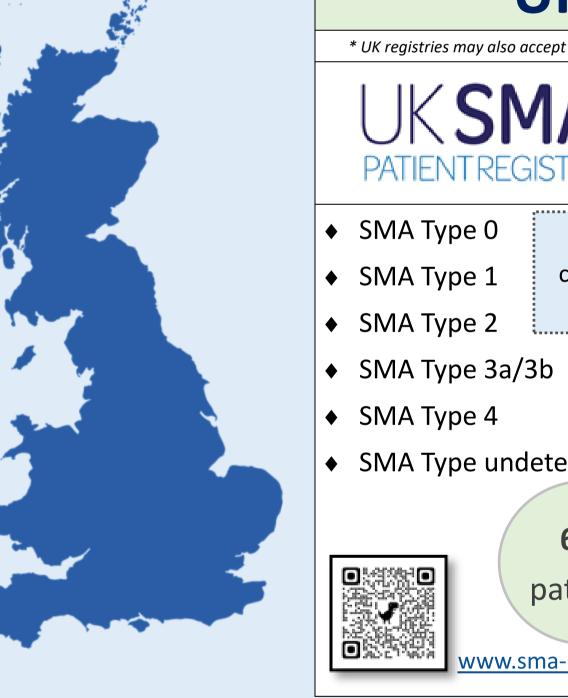


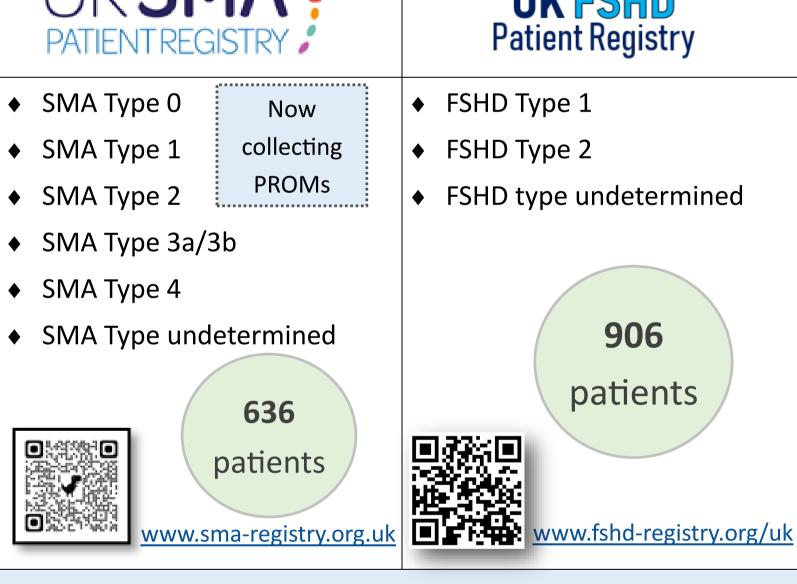
The John Walton Muscular Dystrophy Research Centre (JWMDRC) manages seven national and international patient registries comprising over 4,300 patients. All registries are affiliated with the global neuromuscular network TREAT-NMD and conform to their internationally standardised core datasets, and



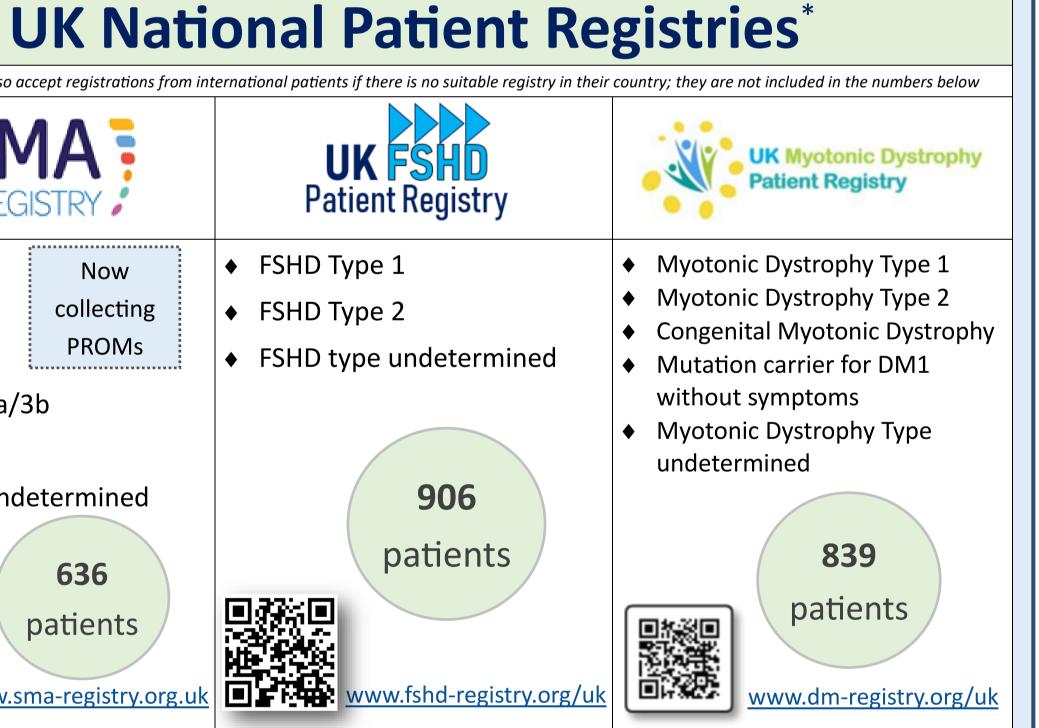
the three national registries contribute data to the TREAT-NMD Global Registries Network (TGDOC).







International Patient Registries



Plans are underway to further extend the registries' research support capabilities through data linkage to the Newcastle Research Biobank for Rare and Neuromuscular Diseases and other initiatives. Work to establish the registries as a Newcastle University Core Research Facility will improve the sustainability and visibility of the registries and increase income generation potential. Future plans also include the development of the registries as Clinical Trial Recruitment Hubs, to support recruitment and equity of access in neuromuscular research, and the use of registry data in support of regulatory post-

marketing surveillance of new

therapies.





Patient numbers correct as of 24/03/2023

GLOBAL FKRP REGISTRY

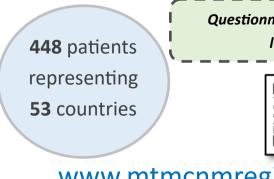
- ♦ Limb-girdle muscular dystrophy type R9 or 2I (LGMDR9 or LGMD2I)
- ◆ Congenital Muscular Dystrophy Type 1C (MDC1C)
- ♦ Muscle Eye Brain Disease
- ♦ Walker-Warburg Syndrome
 - 916 patients representing

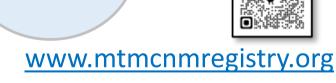
50 countries

www.fkrp-registry.org



- Centronuclear Myopathy X-linked myotubular myopathy
- (XLMTM) ◆ Centronuclear myopathy (CNM)X-
- linked myotubular myopathy (XLMTM)
- ◆ Centronuclear myopathy (CNM)
- ◆ Female carriers of XLMTM
- Deceased MTM or CNM patients









- Ullrich congenital muscular
- dystrophy (UCMD)
- Bethlem myopathy Bethlem/Ullrich intermediate



www.collagen6.org



- ♦ GNE myopathy ♦ Hereditary inclusion body
- myopathy (HIBM)
- ♦ Nonaka myopathy
- Distal myopathy with rimmed vacuoles (DMRV)

424 patients representing **37** countries



Special thanks to our funders:

