Updates and Data Highlights from the Global Registry for COL6-related Dystrophies

An international research database to accelerate the pace of research and treatment.

McDonald S, Pangou I, Walker H, Murphy L, Allamand V, Aimes C, Alvarez R, Dziewczapolski G, Boddy H, Deconinck N, Ferré X, McAlister B, Mejat A, Sarkozy A, Marini-Bettolo C, Volker S.

Global Registry for COL6-related dystrophies

Find out more: **www.collagen6.org**
Contact us: **collagen6registry@newcastle.ac.uk**

WHAT IS THE REGISTRY?

The Global Registry for COL6-related dystrophies (www.collagen6.org) was launched in August 2019 with English as a first language for navigation. We have since translated our registry documents into more languages (Italian, Spanish and French) to increase inclusivity and accessibility to a global level.

The Registry is an open-ended research database, collecting clinical and genetic data and has been created as part of a project funded by the Collagen VI alliance to increase the trial readiness of this group of conditions.

How does it work: Participants log in to complete the questionnaire, upload genetic or biopsy reports, and nominate their doctor to provide some clinical data on their behalf. They are asked to check and update their information annually. The registry supports dual entry, allowing patients and their doctors to enter data.

The purpose of the registry is to:

- Identify participants for clinical research.
- Understand the epidemiology, genetics, symptoms and severity of the conditions.
- Support existing and future research and inform better standards of care.
- Provide a communications interface between patient and research communities.

Inclusion criteria:

- ✓ Living or deceased individuals diagnosed with a COL6-related dystrophy Bethlem Myopathy, Ullrich Congenital, Muscular Dystrophy or Intermediate form (confirmed via genetic testing or biopsy).

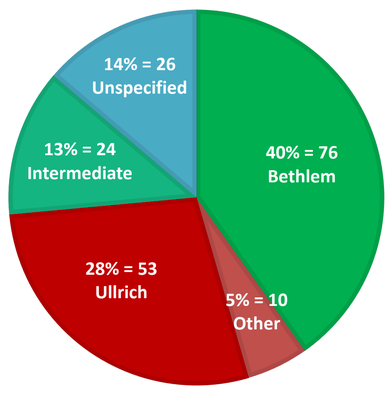
International coverage: Questionnaires are available in English, French, Spanish and Italian but our plans aim to include more languages over the next few years.

COHORT DESCRIPTION

As of November 2022, there are **189 participants** enrolled in the Global COL6 registry, 98 female and 91 male. The mean age of participants is 24 years old.

Registrations are from all over the world, with **32 countries** currently represented, with the greatest registrations coming from USA (29%) and the UK (13%).

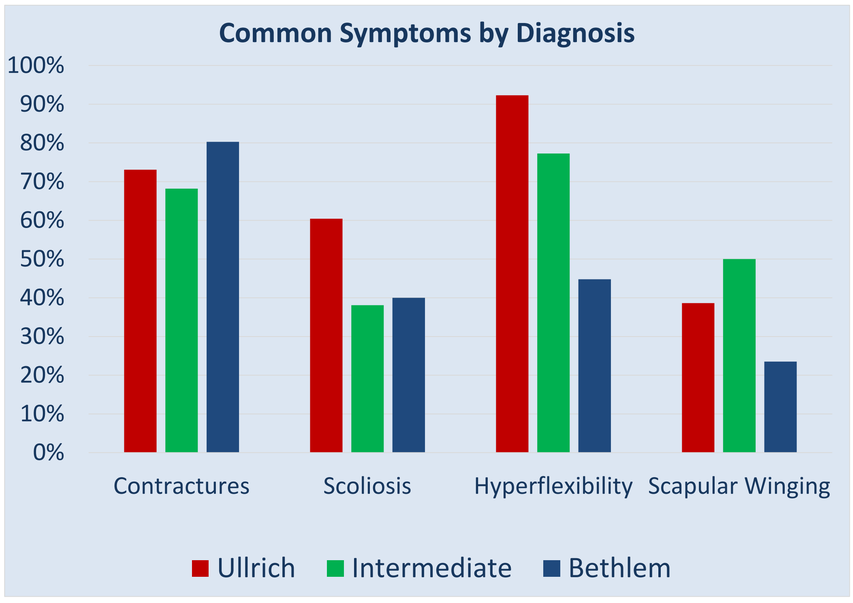
We have begun collection of Genetic Reports from participants, **with 25% (46) participants providing their reports to date.**

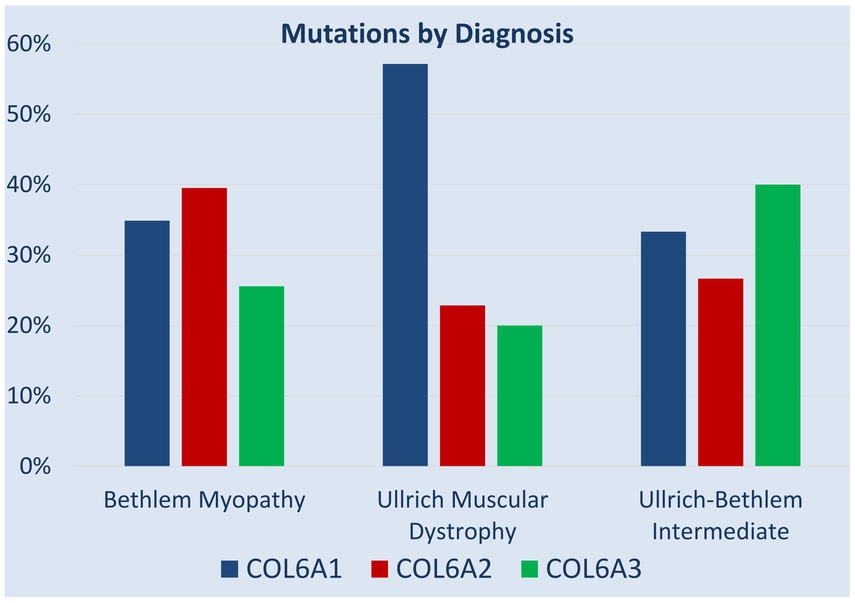


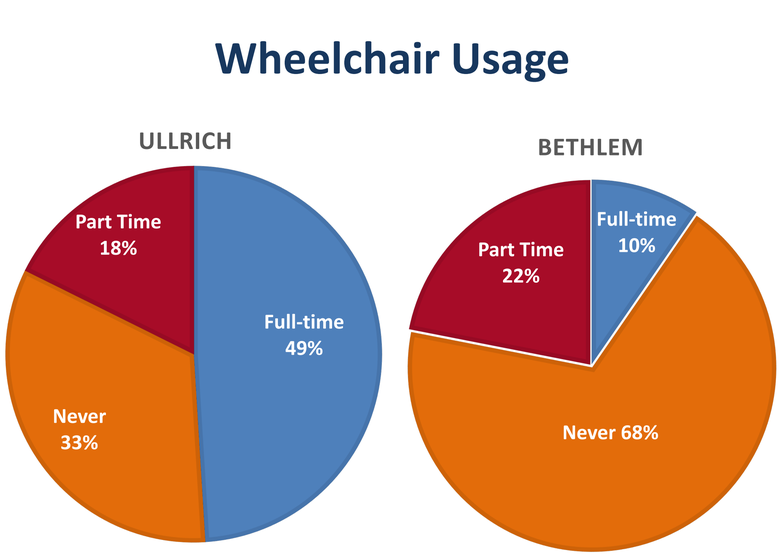
DATASET

Demographics and personal data	Genetic reports
Clinical diagnosis	Neuromuscular examination dates
Skin changes	Motor function
Wheelchair use	Respiratory function and ventilation use
Scoliosis surgery	Unplanned hospitalisations
Inheritance Pattern	Feeding function
Muscle Pain	Family history

CLINICAL FEATURES







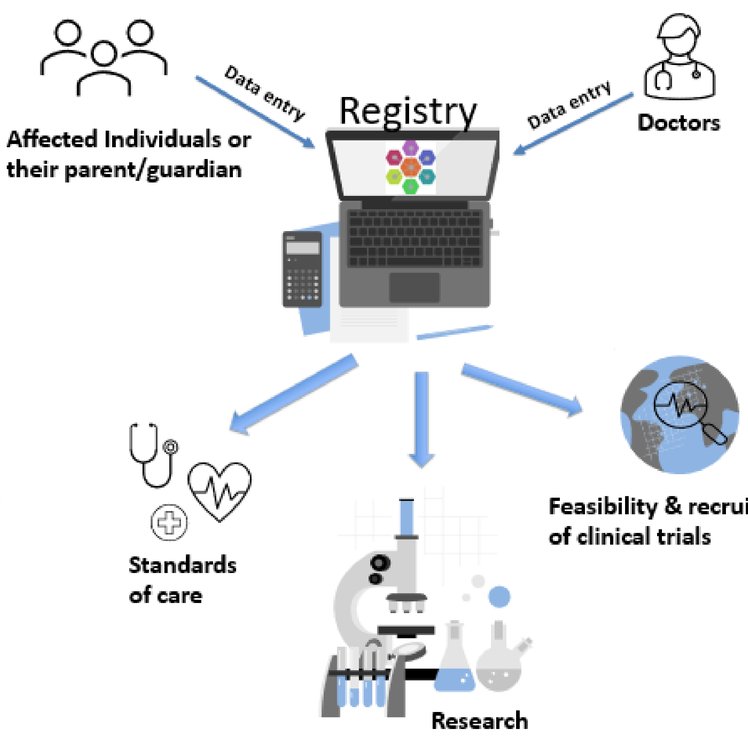
Registry Use and Engagement with TREAT-NMD

The Global Registry for COL6 Related Dystrophies has been involved in a number of enquiries, including surveys to:

- Understand the impact of COVID-19 on patients with neuromuscular disease
- Assess swallowing difficulties in NMD to help optimise UK service delivery
- Understand more about fertility and pregnancy in neuromuscular disease

As an **affiliate of the TREAT-NMD Global Data Oversight Committee**, the registry works hard to harmonise approaches with other neuromuscular registries. To support this, we are planning to implement the **LGMD core dataset** for patients enrolled with a diagnosis of LGMD R22 or D5 in Spring 2023, with the rollout of our updated registry platform version 2. This will also provide a mobile-friendly interface for patients to enrol on their phone.

TREAT-NMD Neuromuscular Network *Member of the TREAT-NMD Neuromuscular Network*



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Registry website
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Muscular Dystrophy UK
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