# Global Registry for COL6-related dystrophies

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Find out more: **www.collagen6.org**
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## BACKGROUND

The Global Registry for COL6-related dystrophies 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 and support research in 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:**

- Support readiness for future clinical trials
- Understand the epidemiology, genetics, and natural history of COL6-related conditions
- Support existing and future research and inform better standards of care.
- Provide a communications interface between patient and research communities.

## COHORT DATA

***Table 1**. Current Participants (23/03/2023*

| | Male | Female | Total |
|---|---|---|---|
| **Patients** | **98** | **96** | **194** |
| **Genetically confirmed patients** | **25** | **36** | **61** |

***Table 2**. Registry Dataset*

| PATIENT REPORTED DATA | |
|---|---|
| Demographics | Genetic reports |
| Clinical diagnosis | Neuromuscular examination dates |
| Skin changes | Motor function |
| Wheelchair use | Respiratory function/ventilation use |
| Scoliosis surgery | Unplanned hospitalisations |
| Quality of Life | Feeding function |
| Muscle Pain | Family history |

| CLINICIAN REPORTED DATA | |
|---|---|
| Ambulatory status | Age of first symptoms |
| Medications | First presenting symptoms |
| Age at diagnosis | Pulmonary Function |
| Clinical & Molecular Diagnosis | Comorbidities |
| Skin biopsy results | Gait and Wheelchair Use |
| Muscle biopsy results | Cognitive function |
| Muscle MRI results | Current Symptoms |

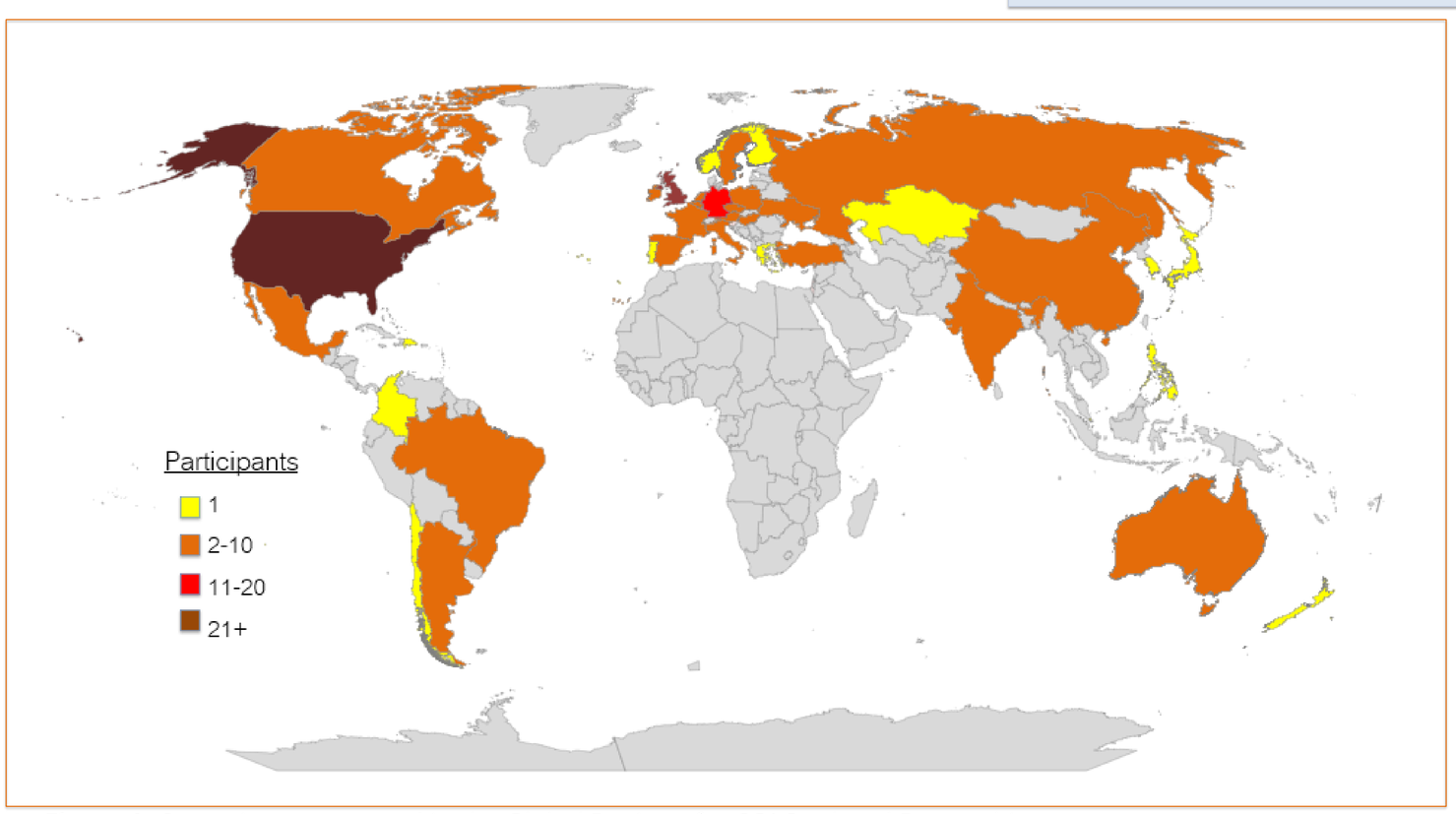


***Figure 1**. Countries represented in the Global Registry for COL6-related Dystrophies*

## FUTURE PLANS

**Dataset Review**

The registry's Steering Committee is comprised of expert clinicians, researchers, affected patients and family members, and representatives from patient advocacy organisations. Their role is to provide guidance on the direction of the registry. Presently, the steering committee is reviewing the current dataset (**Table 2**), to ensure it remains as useful and representative as possible

**Translations**

The registry questionnaires, information sheets and consent forms are available in 6 languages – English, Spanish, French, Italian, Swedish and Polish. Work is ongoing to enable participants to access the website in their preferred language, starting with Spanish, scheduled for June 2023.

**Reaching further participants**

A coordinated approach by the John Walton Centre's international registries is being planned for Central and South America. New connections and collaborations have also been established with clinicians in South Korea, Japan and New Zealand.

## REGISTRY PARTICIPANTS

There are currently 194 participants in the registry. Genetic reports confirming diagnosis and affected gene have been received by 31% of participants, and continues to increase. **39 countries** are represented on the registry, with the majority of registrations coming from Germany (6%), the United Kingdom (12%) and the USA (24%).
COL6A1 is the most commonly reported affected gene (33%), and this is supported by the results of confirmed genetic diagnoses (50%).

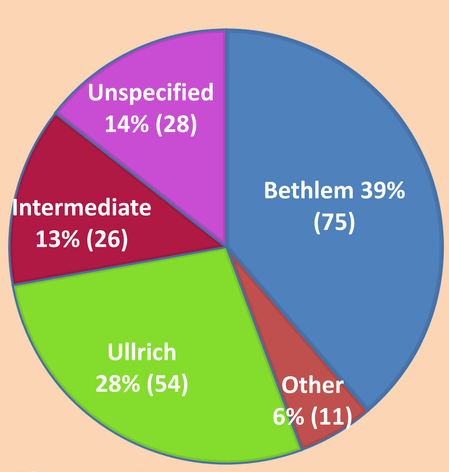


***Figure 2**. Patient reported diagnosis*

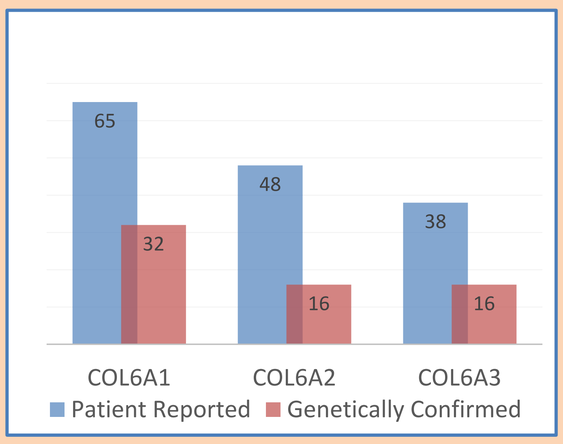


***Figure 3**. Affected gene*

## TREAT-NMD Affiliation

As an affiliate of the TREAT-NMD Global Data Oversight Committee, the registry works hard to harmonise approaches with other neuromuscular registries.

By collecting and analysing data from patients around the world, TREAT-NMD is able to support registries in tracking the progression of patients over time, and facilitate highly powered research studies.

To support this work, we are working to implement TREAT-NMD's LGMD core dataset for patients enrolled with a diagnosis of LGMD R22 or D5 in 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.

**LGMD Core Minimum Dataset**

TREAT-NMD Neuromuscular Network

**Sam McDonald**
Registry Coordinator

**Professor Volker Straub**
Principal Investigator

**Provide data for your patients**

**Registry Website**
www.collagen6.org

*With thanks to MDUK*

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