

Global Registry for COL6-related dystrophies

Dr Alison Blain*, Prof. Volker Straub*

*The John Walton Muscular Dystrophy Research Centre, Institute of Genetic Medicine, International Centre for Life, Newcastle upon Tyne NE1 3BZ.

Collagen6registry@ncl.ac.uk

The **Global Registry for COL6-related dystrophies** (www.collagen6.org) was launched in English in August 2018 as part of a project funded by the **Collagen VI alliance** to increase the **trial readiness** of this group of conditions.



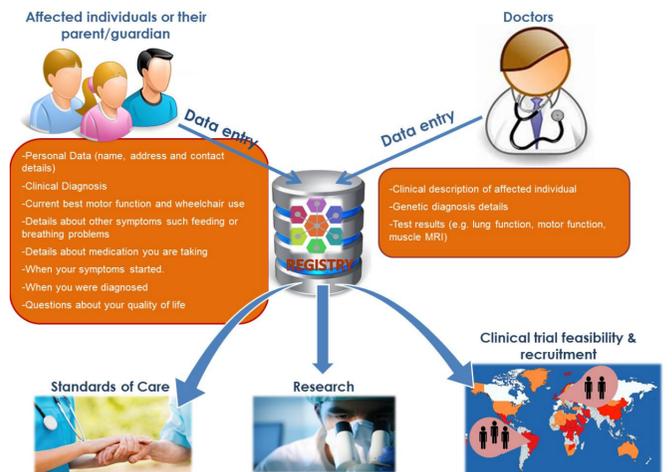
The registry aims to:

- Help identify patients for relevant **clinical trials** as they become available.
- **Encourage further research** into Collagen 6-related dystrophies.
- Provide researchers with answers to specific questions to **support research**.
- Assist doctors and other health professionals by providing them with up-to-date information on managing Collagen 6-related dystrophies, to help them deliver **better standards of care** for their patients.

And welcomes the registration of:

- All patients, with a diagnosis of **Bethlem Myopathy**, **Ullrich Congenital Muscular Dystrophy (UCMD)** or **Intermediate form** of these conditions.

The registry is dual entry, allowing patients and their doctors to enter data



The registry is currently being translated into a number of languages: Italian, Spanish, German, Polish, Dutch, Swedish, French.

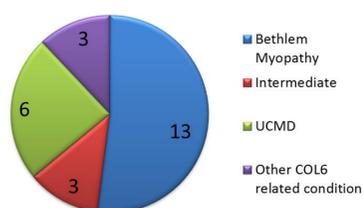


The registry has the following approvals: HRA Research Ethics Committee, Newcastle University Ethics, Newcastle upon Tyne Hospitals Trust (NuTH) R&D, NuTH Caldicott Guardian approval, Royal Children's Hospital Melbourne Research Ethics Committee (HREC), Governance authorisation at Melbourne Children's Campus.

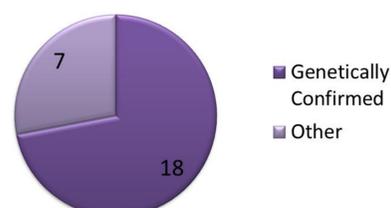
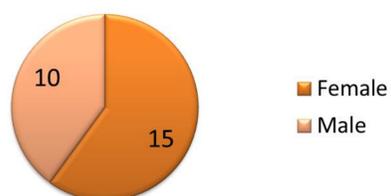
Participant Demographics (as of Feb 2019)



Clinical Diagnosis

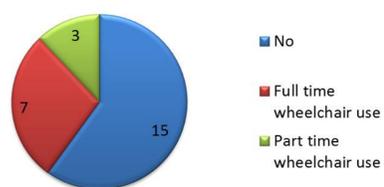


Sex



Selected clinical features

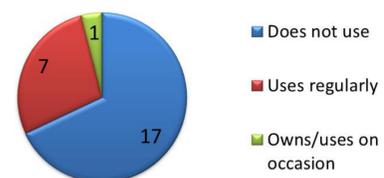
Wheelchair use



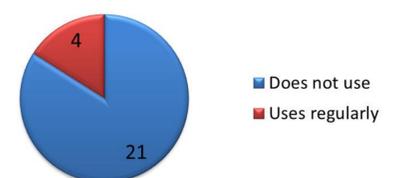
Current best motor function



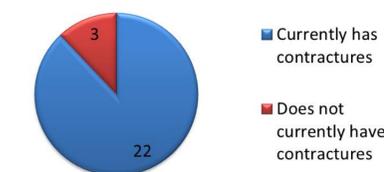
Non-invasive ventilation use



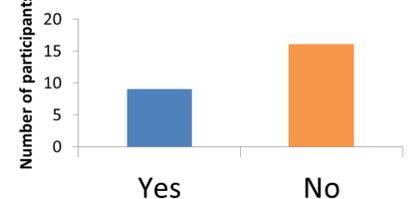
Invasive ventilation use



Contractures



Surgery to correct contractures?



Unplanned hospital admissions

1 out of 25 registry participants have had an unplanned hospital admission in the past year.

The registry is a flexible tool for trial readiness and research in the COL6-related dystrophies.

If you are a clinician or patient who would like to take part, or a researcher who would like to learn more about how registries can support research, please contact the registry curator at: collagen6registry@ncl.ac.uk