Global Registry for COL6-related dystrophies

INFORMATION FOR PARENTS

Principal Investigator: Volker Straub, a Professor of Medicine at Newcastle University, UK

Should you have questions relating to the registry, you can contact either your child's local doctor or the registry curator Alison Blain, a research associate at Newcastle University (you can find her contact information below).

Before you agree to register your child in the Global Registry for COL6-related dystrophies, it is important that you understand what is involved and what will be done with the information that you provide. This form contains the answers to the questions that you might have. At the end of the form there is a checkbox for you to click on to confirm that you have read the information. If you have any questions after reading this form, please contact us before continuing.

What is a patient registry and why do we want to create one?

Scientific advances over recent years have led to substantial changes in the treatment of many disorders. New therapeutic strategies are being developed and, for some of these treatments, plans for large studies involving patients from more than one country are already in place.

Several new therapeutic strategies for neuromuscular disorders target specific gene defects. When a scientific study or clinical trial is being planned, it is very important that patients suitable for that trial can be found and contacted quickly. The best way to ensure that this happens is to make sure that patients' details are all collected in a single database or "registry" that contains all the information that researchers will need, including each patient's particular genetic defect and other key information about that disorder.

Newcastle University has created an international registry for COL6-related dystrophies, which means that all patients who register will be contacted if their profile fits the requirements for a particular study or clinical trial. In addition the registry will help researchers answer questions such as how common disorders like COL6-related dystrophies are distributed internationally, and will support other activities to improve patient care and establish a good standard of care worldwide. The registry team is based at the John Walton Muscular Dystrophy Research Centre at Newcastle University, UK, and is part of the TREAT- NMD alliance (http://www.treat-nmd.eu/) global network of registries. We will always notify you about significant changes to the database management and team.

Who is funding the registry?

The project is funded by Muscular Dystrophy UK (MDUK), in partnership with the Collagen VI alliance, have provided funding for the project for 3 years however we hope to keep the registry running indefinitely with new funding. No additional payments will be received by Straub or other members of the registry team for adding your child's details to the database.

Whose data are we collecting in this registry?

The Global Registry for COL6-related dystrophies is for patients affected by Ullrich congenital muscular dystrophy (UCMD), Bethlem myopathy (BM) and intermediate forms of these conditions, caused by mutations in the Collagen 6 genes. The Global Registry for COL6-related dystrophies is primarily designed to register patients who might be suitable for participation in future research studies or clinical trials of new therapies, and to help the researchers find the best way of caring for patients with UCMD/BM and other Collagen 6 -related conditions. This registry is intended for patients currently living with the condition, as well as those who are deceased but had a genetically confirmed diagnosis.

What does my child have to do?

If you agree to your child taking part in this project, then you should read this information and tick the checkbox at the end. You will then be asked to complete a consent form, this confirms that you agree to your child participating. If your child is aged 11-15, they will also need to complete an assent form (when your child turns 16 we will need them to complete a consent form but you will be contacted about this). You should then complete the online registration questionnaire, in which we ask you for some of your child's personal data and some information about their condition. **Since the COL6-related dystrophies can vary in how they affect an individual, the questions are designed to cover a whole range of symptoms which your child may, or may not have. It should be noted that your child may never suffer from some of the symptoms which are asked about however, we understand that answering these questions may be emotional. Please feel free to take a break or discontinue filling out the questionnaire.**

As part of the questionnaire, you will be asked to indicate who your child's neuromuscular specialist clinician is so that they can be contacted by the registry curator to complete the professional part of the questionnaire and to verify your database entry. If your child has had a Muscle MRI as part of their normal care, we will ask if you consent to their doctor submitting their scan data anonymously to the MYO-MRI scanbank image repository. Their MRI scan data will be identified in the scanbank using their registry unique code.

The information that you and your child's clinician provides will be entered into an international registry which is supervised by the registry steering committee. Details of current membership of the steering committee and the role of the steering committee can be found at: https ://collagen6.org/steering-committee/

How can I update my child's data if something has changed?

To make sure that the data in the registry is correct and up-to-date, it is essential that we update it regularly. To do this, we will send you emails once a year asking you to tell us about any changes in your child's medical condition. We also ask you to inform us if there are any major changes in your child's details that might occur in the period between updates, for example a change of address or the loss of ambulation.

Who will have access to my child's data?

Staff in charge of the registry will have access to your child's data to obtain information necessary to a project and are able to contact you, for example to inform you about an upcoming clinical trial. Also the doctor that you choose during registration will have access to your child's data, this is necessary because he/she will fill out the second part of the registration questionnaire.

How will my child be identified in the registry?

Your child's personal details (name, address etc.) have to be stored in the registry so that we can contact you if we need to inform you about possible clinical trials or research studies or anything else that might be relevant to your child's disorder. This data will be stored in a secure manner and your child's records will be assigned a unique code. Your child's data will

only be identified by this unique code when data is exported from the registry for analysis.

Only the person in charge of the registry (Volker Straub) and the registry curator appointed by him will be able to access your child's personal details.

Will information about my child be kept confidential?

Creating a registry requires the existence of a file containing a patient's personal and medical data. Your child's data will be encrypted and stored securely within the European Union, under the responsibility of Volker Straub. Your data will be stored indefinitely and treated with confidentiality. For more details about the database and how it is hosted see: https://collagen6.org/the-database/

If you would like more information about how we manage personal data more generally, including your rights under law, and the contact details of the University's Data Protection Officer, please see our website: http://www.ncl.ac.uk/data.protection/PrivacyNotice

How will my data be used?

When planning a scientific study or clinical trial, researchers can submit questions about the data contained in the registry to the registry curator and if their 'enquiry' is approved by the registry steering committee, they will be given anonymised aggregate data in the form of a written report.

We will not give your child's personal information to clinical or scientific researchers. If we are contacted by a researcher who is recruiting for a clinical trial, we will contact you if we think your child may potentially fit the criteria for the study. If you are interested in the information that you receive about a particular clinical trial, you will be given further information about how you can contact the researchers running the trial. If you and your child decide that they would like to take part in the trial, you will need to review and sign a separate consent form**. You are completely free to make your own decision about any trial we inform you about**. If you decide that you do not wish your child to take part in a particular trial, their data will still be kept in the registry and we will continue to inform you about other trials unless you tell us not to. Please note that if we tell you about the existence of a trial, this does not imply that we endorse it.

Your child's data will not be made available to employers, government organisations, insurance companies or educational institutions, nor to other members of your family. If we publish any research or other documents based on the data from the registry, this research will never identify your child by name.

Data linkage to support research

The registry is working to anonymously link data in the registry to data held in other research data repositories (namely, the **Neuromuscular Biobank** based at Newcastle University, the **ScanBank MRI repository**, the **Congenital Muscular Dystrophy International Registry (CMDIR)** and the RD-Connect **Genome-Phenome Analysis Platform**). The reasons we would like to do this are three-fold; it enhances the information available to researchers when accessing biomaterials, it supports research aiming to understand the relationship between the genetic variants present and the clinical presentation (genotype-phenotype relationships), and facilitates the validation of imaging as a potential biomarker for use in future trials. Data would be linked via a unique identifier and in a way which ensures confidentiality is preserved.

How will my child benefit from registering?

The registry is intended as a public service for the benefit of patients living with UCMD, BM and other Collagen 6-related conditions. You will not receive any payment or any other financial benefit as a result of submitting your child's data to the registry. The results of research facilitated by the registry may be patentable or may have commercial potential. However,

you will not receive patent rights and will not receive financial benefits from future commercial development. Nevertheless, there are other benefits from participating, including the following: We will inform you if (on the basis of the information that you and your doctor provide) your child might be a suitable candidate for a certain clinical trial. We will also inform you if we receive any new information on your child's disorder which might be of interest to you - for example if we find better ways for caring for patients with UCMD, BM and other Collagen 6-related conditions. The data collected might also provide benefits for other patients with your child's disorder, for example by revealing statistics on how many people worldwide have the same condition, or providing information for researchers interested in the best standards of care for your child's disorder.

I want my child to be involved in a clinical trial – is this guaranteed if we register?

Although one of the main aims of this registry is to make it easier for patients to be recruited for trials or studies, there is no guarantee that registering your child's details will ensure that they will be involved in a clinical trial or research study. If you are interested in receiving details of trials or studies that your child may be eligible for, please select this option in the questionnaire. However, it is important that you understand that even if the registry curator believes that your child might be eligible for that trial or study, based on the data about your child stored in the registry, it is possible that at a later date it will turn out that your child does not meet the inclusion criteria for the trial/study after all.

I do not want my child to take part in a clinical trial or research study. Should we still register?

We hope that you will be interested in registering even if your child will not take part in a clinical trial or research study. Their information will still be useful to researchers who are trying to find out more information about patients living with Collagen 6-related conditions, and we will still provide you with other information that might be relevant to your child's disorder. It is up to you whether or not you act on any information we give you about clinical trials or research studies.

Does my child have to participate in the registry? Can we change our mind?

Your child's participation in this project is completely voluntary. You can decide for your child not to participate in this registry without having to give any reason. If you wish for your child to join this registry, you have the right to access your child's data and to view, rectify or update it at any time. Should you wish to withdraw your child's from the registry, you will be free to do so at any time without having to provide any explanation and without consequence on their treatment or the quality of care that will be provided to them. If you wish to withdraw your child, you will need to get in touch with the staff in charge of the registry. Contact details are provided below.

Who should I contact if I have any other questions?

If you would like any additional information about the registry, if you need to tell us about any changes in your child's data, or if you wish to withdraw your child and/or your child's data from the registry, please contact:

The registry staff using this email address: collagen6registry@ncl.ac.uk
Phone number: 0191 241 8640

Or the registry principal investigator:
Volker Straub:
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This study was approved by the Faculty of Medical Sciences Research Ethics Committee, part of Newcastle University's Research Ethics Committee. This committee contains members who are internal

to the Faculty, as well as one external member. This study was reviewed by members of the committee, who must provide impartial advice and avoid significant conflicts of interests.