



## Who should register?

Patients with limb girdle muscular dystrophy 2I (LGMD2I) or congenital muscular dystrophy 1C (MDC1C) or another condition with a confirmed mutation in the FKRP gene can register. Young people under the age of 16 years must be registered by their parents or guardians. This registry is for all patients worldwide.

If you have a diagnosis of LGMD or CMD but aren't sure which kind it is or whether you have had a genetic test to confirm the mutation, you can ask your doctor for help or contact us by email or post at the address overleaf.

Since patients with FKRP mutations are rare, every single person counts!

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## How do I register?

Registration is voluntary and is done by the patients themselves or by their parents/guardians. Patients can register online (over the internet), which allows them to view and update their data at any time.

Register online at:

[www.FKRP-registry.org](http://www.FKRP-registry.org)

Information that you enter online is encrypted while being transferred, so that it cannot be intercepted.

If you cannot register online, contact us at the address overleaf.

## Why should I register?

There are several reasons for patients to register:

- Registries facilitate patient recruitment for clinical trials and registered patients may be able to participate in trials more easily.
- Registries aim to accelerate research into new therapies for LGMD2I, MDC1C and other FKRP-related conditions.
- Registered patients are kept informed about research results, such as new treatments for these conditions, as well as about TREAT-NMD activities.
- Registries help specialists gain more knowledge about the prevalence and natural history of LGMD2I, MDC1C and other FKRP-related conditions.
- Registries may help achieve equal care for all patients with these conditions worldwide.
- Registries may help raise the urgently needed funds for further research.

## UK contact point:

FKRP patient registry, TREAT-NMD office  
Institute of Human Genetics, Newcastle University  
International Centre for Life  
Newcastle upon Tyne, NE1 3BZ

Local contact email: [UK@FKRP-registry.org](mailto:UK@FKRP-registry.org)

## International registry principal investigator:

Dr Maggie Walter  
Friedrich-Baur Institute, Department of Neurology  
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# Global FKRP Patient Registry

An opportunity for patients with LGMD2I, MDC1C and other FKRP-related conditions to participate in clinical trials and obtain the best possible care



Register online at:  
[www.FKRP-registry.org](http://www.FKRP-registry.org)

## What is a patient registry?

A patient registry collects information about patients who are affected by a particular condition. The international FKRP patient registry collects genetic and clinical data about patients affected by a mutation in the FKRP gene. Mutations in this gene cause one of the forms of limb girdle muscular dystrophy (LGMD) known as LGMD2I, and one of the forms of congenital muscular dystrophy (CMD) known as MDC1C. In very rare instances, FKRP mutations can also cause the conditions known as Muscle Eye Brain disease and Walker-Warburg syndrome. LGMD2I is by far the most common of the disorders caused by an FKRP mutation. For research into treatments for all of these conditions, it is important that researchers have precise information about the genetic mutation that is responsible. In anonymous form, valuable medical data from the registry will be made available to selected researchers around the world, thereby accelerating the research into conditions caused by FKRP mutations and their possible treatments.

Additionally, with the advent of clinical trials for some neuromuscular conditions, patient registries mean that patients who may be eligible for certain clinical trials are readily identifiable. The registries contain accurate and updated information about the patients' genetic mutations and their clinical condition. This information is provided both by the patients themselves and by the professionals involved in the patient's care, after the patient has given full consent. Because FKRP mutations are rare conditions, without a patient registry to gather patient details from all FKRP patients worldwide in one place, finding enough patients for a meaningful trial can take years, and this delays the testing of potential therapies.

## What data is stored in the registry?

The global FKRP registry contains patients' personal details, such as name, address, date of birth and gender, so that they can be identified and contacted by the registry staff where appropriate. It also contains certain items of clinical and genetic information.

Doctors and scientists can access this medical data and use it for their research into the conditions. They can also use the registry to identify patients for clinical trials, but patients' personal details are protected and only known to the registry staff. All the data is stored on secured computers accessible only by selected personnel.



## What is TREAT-NMD?

It is an EU funded network bringing together people with neuromuscular disorders and specialists (scientists, healthcare professionals and pharmaceutical companies) working on treatments for these conditions.

- The network aims to help speed up the research that is being done into these conditions so that the most promising new therapies can move more quickly from being an idea in a laboratory to a real treatment for patients.
- It involves many different people – from the geneticists who look for the problems in the genes, to the laboratory scientists who study the body cells involved, to the doctors who treat the patients, and finally the patients themselves – and by improving the way all these people work together across Europe and worldwide, it aims to make real progress in a short time.

The FKRP patient registry is just one of TREAT-NMD's activities aiming to benefit patients worldwide. The global TREAT-NMD registry for FKRP collects medical information on FKRP patients from all over the world, which means that should a potential treatment become available, eligible patients can be contacted easily. More information about the TREAT-NMD network and its activities is available at [www.treat-nmd.eu](http://www.treat-nmd.eu).



"...we have lived through the despair of an initial incorrect diagnosis, the diagnostic odyssey with multiple inconclusive doctor visits, the joy of seeing our disabled daughter surmount challenges and bravely with spunk face each new day and the desire to forget the diagnosis and lead as normal a life as possible. Yet, we also live with suppressed fear, waiting for the day that she becomes weaker and the uncertainty of her prognosis. A registry signifies a great leap forward towards defining who our children and adults are and how best medical practices can help maintain their strength, bolstering their fragility, allowing them to reach their fullest potential. A registry is a harbinger of a change in the medical cultural mindset interacting with the muscular dystrophies as a treatable disease. A registry raises the flag of hope and carries the promise of confident minds looking towards a future that may one day hold clinical trials and therapies that slow disease progression in our children and affected adults."

Anne Rutkowski and Joe Pinkelman, parents of Maia, 10 years old. Anne is co-founder and vice-chairman of Cure CMD.



"A registry for persons affected by mutations in the FKRP gene, will generate important knowledge about the natural history of the disease, which will directly impact on the clinical follow-up of patients. Molecular therapies for inherited muscle diseases, including FKRP, are emerging, and a registry will facilitate recruitment to such studies, which typically involve multiple centers in different countries. For the patients, participation will provide them with up to date information about treatment of their condition, and the possibility to join treatment trials. I therefore endorse the registry, and strongly encourage affected individuals to register."

John Vissing, Professor of Neurology and director of the Neuromuscular Clinic and Research Unit, Denmark

"It is a wonderful idea. Knowing that there is all this work going on gives us hope for the future."

Carol Turnbull and Susan Mitchell, sisters with LGMD2I.

