

## **CMD Research Resource Guide: Frequently Asked Questions on the Congenital Muscle Disease International Registry (CMDIR), BioBank, and Tissue Repository**

### **Who can participate in the Registry, BioBank and Tissue Repository?**

Individuals with the following diagnoses are encouraged to participate. Genetic confirmation of disease is not required for participation. If you do not yet have genetic confirmation of disease, CMDIR counselors can help discuss genetic testing with you.

### **Congenital muscular dystrophy subtypes included in the CMDIR**

- Alpha 7 Integrin Related Myopathy
- Alpha 9 Integrin Related Myopathy
- Collagen VI Related Myopathy (Ullrich through Bethlem CMD)
- Alpha-Dystroglycan Related Muscular Dystrophy (Dystroglycanopathy, WWS, MEB, Fukuyama, FKR, LGMD2I, LGMD2K, LGMD2M, LGMD2N, LGMD2O)
- Choline Kinase B Receptor
- LAMA2 Related Muscular Dystrophy (Laminin Alpha 2 related dystrophy/MDC1A/Merosin deficient)
- LMNA Related Muscular Dystrophy (Laminopathy/LaminA/C, L-CMD, Emery Dreifuss muscular dystrophy)
- SEPN1 Related Myopathy (Rigid Spine Muscular Dystrophy/RSMD1, Congenital Fiber Type Disproportion, Mallory Weiss Body Desmin, Multi-minicore Myopathy)
- SYNE1 (Nesprin Related Muscular Dystrophy)
- Telethonin Related Muscular Dystrophy (TCAP/Titin-Cap)
- CMD Undiagnosed (including Merosin Positive)

### **Congenital Myopathy subtypes included in CMDIR**

- Actin aggregation myopathy
- Cap disease
- Central core disease
- Centronuclear myopathy
- Congenital fiber type disproportion
- Core rod myopathy
- Hyaline body myopathy
- Multiminicore myopathy
- Myofibrillar myopathy
- Myotubular myopathy
- Nemaline myopathy
- Reducing body myopathy
- Spheroid body myopathy
- Tubular aggregate myopathy
- Zebra body disease myopathy
- Congenital Myopathy with unknown subtype

### **Congenital Myasthenic Syndrome (all subtypes included in CMDIR)**

# CMDIR

**Congenital Muscle Disease International Registry**

*Leading the Way to a Treatment and Cure*

## **What is a patient registry?**

A registry is like a phonebook that stores names, phone numbers and information about people. A patient registry is a database of medical information gathered to:

- Count the number of people with a given disease
- Raise awareness
- Support research working towards a cure
- See how the disease affects people differently
- Connect researchers and families for treatment
- Help doctors make the best health plan for people with muscle diseases

## **About the Congenital Muscle Disease International Registry**

The Congenital Muscle Disease International Registry (CMDIR) was created to identify the global congenital muscle disease population and includes individuals with congenital myopathies (all subtypes), congenital muscular dystrophies (all subtypes), congenital myasthenic syndromes, and the limb girdle spectrum. The CMDIR collects demographic information, disease symptom information, and medical records. The CMDIR interacts with the CMD BioBank and CMD Tissue Repository to provide medical information to accompany banked blood or tissue samples. Information like highest motor function, whether or not the donor uses breathing support or is taking any medications is useful to scientists working with donated blood or tissue samples.

## **Why should I register in the CMDIR?**

Simply put, we will not be successful in finding effective treatments, or eventually cures, unless we know who the affected individuals are, what their diagnoses are, and how the disease is affecting them.

## **What is a BioBank/Repository?**

BioBanks and repositories are places to store samples from the body. The Congenital Muscle Disease (CMD) Tissue Repository collects, stores, and redistributes tissue samples from surgical and autopsy tissue to provide a central resource for scientists who study muscle disease. The CMD Tissue Repository also can establish cell lines from fresh blood or tissue, a portion of which are then sent to the CMD BioBank within the NIGMS Repository at the Coriell Institute for Medical Research.

## **About the Congenital Muscle Disease Tissue Repository**

The CMD Tissue Repository is a place for scientists to store body tissues collected during surgeries or autopsies that can be used for research to help understand more about the disease, and to help develop treatments and cures for congenital muscle diseases. This repository is located at the Medical College of Wisconsin (USA).

## **About the Congenital Muscle Disease BioBank**

# CMDIR

**Congenital Muscle Disease International Registry**

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The CMD BioBank is located within the NIGMS Repository at the Coriell Institute for Medical Research in Camden, New Jersey. The purpose of the CMD BioBank is to accelerate research on the congenital muscle diseases by banking cell lines from patients with muscle disease. You can donate directly to the CMD BioBank or through a combined donation of blood and tissue to the CMD Tissue Repository, and the consent process will be initially managed through the same people. Coriell uses samples donated to the CMD BioBank to make 4 important types of resources for scientists: fibroblast cell lines, lymphoblast cell lines, buffy coats and iPS cell lines.

### **Why should I donate a sample to the CMD Tissue Repository and/or CMD BioBank?**

Because the congenital muscle diseases are rare, it is often very difficult for scientists to obtain samples for their research. That is what makes your donation such an important gift. Scientists need tissue to understand the disease better and work towards a treatment. Having human tissue allows scientists to gain greater insight into how the different organs are potentially affected (heart, brain, intestine, nerve, liver), what happens in these tissues over time, and how cells from other areas of the body (heart, brain, intestine, nerve, liver) might respond to drug compounds.

### **Who can donate a sample to the Tissue Repository and/or CMD BioBank?**

People who have congenital myopathy (all subtypes), congenital myasthenic syndrome and congenital muscular dystrophy (all subtypes) through the late onset spectrum can donate to the CMD Tissue Repository. Genetic diagnosis is not required to participate.

The CMD BioBank collects samples for the establishment of cell lines from affected individuals, their parents and their siblings. Knowledge of the genetic cause is not necessary to donate a sample. Parent and sibling samples are only collected if the affected individual has also donated a sample to the BioBank. Obtaining blood samples from family members and the affected individual makes it possible to perform “epigenetic studies” in the future. These studies look at disease-modifying genes. Understanding disease-modifying genes can help us learn why some forms of congenital muscle disease are milder for some people and more severe for others.

### **What happens to my sample once it is donated?**

Tissue that is donated to the CMD Tissue Repository will be safely stored and scientists will be able to request this tissue for their research studies. Portions of your donated tissue will be distributed for projects that are deemed worthwhile. Information related to the sample, such as the severity of your disease and any known genetic abnormalities, may be shared with researchers, but your personal identifying information will not be shared. If your donation to the CMD Tissue Repository includes blood or tissue that can be used to establish cell lines, then we will attempt to establish these cell lines and will ship a portion of the cells to the CMD BioBank.

Blood that is donated directly to the CMD BioBank at Coriell will be spun down and cells will be removed. These cells will be encouraged to grow and multiply. Once the cells are thriving, they will be put into separate test tubes and stored in a freezer. The cell lines, called lymphoblast cell lines, will then be placed in the NIGMS Repository catalogue to allow scientists to obtain the cells for research. A similar process is used for the skin cells. If you are able to donate 2 tubes of blood, your second tube will be spun down to create a “buffy coat”. The buffy coat contains white blood cells, which are stored, without creating cell lines for future genetic studies.

### **How will my sample be used by scientists?**

Samples donated to the CMD BioBank can be used by scientists for a variety of purposes, including: development of therapies, discovery of disease genes and their function, further study of known genes/gene expression, and development of new genetic tests.

### **Do I have access to the tissue I donate?**

You have the opportunity to discontinue your enrollment in the CMD Tissue Repository at any time and the information and tissue banked on-site will be destroyed. Tissue and information that has already been distributed cannot be reclaimed. Cell lines established at the CMD BioBank at the Coriell Institute for Medical Research are de-identified before banking there, and are not able to be reclaimed or destroyed after a patient has discontinued their participation in the CMD BioBank.

### **How does the Tissue Repository maintain my privacy and confidentiality?**

The CMD Tissue Repository takes the privacy and confidentiality of each donor very seriously. We use the following measures to protect the privacy of donors:

1. Assign a code number to each sample that is linked to your profile in the CMD International Registry
2. Remove donor’s name and any personally identifying information
3. Follow strict guidelines that forbid us from distributing any personally identifying information to scientists getting the samples

The CMD Tissue Repository will require access to some identifying information, such as the patient’s name, birth date, and phone number, in order to perform the consenting process and appropriately organize the banking of the tissue. This information will not be shared.

### **Does it cost money to donate a sample?**

For tissue and blood donations to the CMD Tissue Repository, the Repository will cover the costs of shipping, and has funds available to cover the costs associated with tissue collection on a case-by-case basis. Our goal is to prevent any costs associated with your tissue donation.

For blood specimens that are directly shipped to the CMD BioBank at Coriell, Coriell will pay for the cost of shipping. Coriell can reimburse you up to \$40 for the cost of a blood draw. It may be simplest to collect the sample during a regular doctor's visit or when blood is being drawn for another reason. Coriell cannot pay for any fees that come from having a skin biopsy, so it's best to have skin biopsies collected during other scheduled surgeries to avoid additional costs.

**How do I donate a sample to the CMD Tissue Repository and/or CMD BioBank?**

There are a few ways in which a donation can be made. 1) The first is through a surgical procedure such as cardiac transplant, scoliosis surgery, contracture release, or muscle biopsy. Arrangements can be made in advance for a tissue sample to be obtained during this surgery from discarded tissue (not clinically useful) and then be made available to scientists for study. This would pose no extra risk to you or your family member other than the known risks associated with the surgical procedure. 2) There is also the option of donating blood for the establishment of cell lines, which can either be done during a medical procedure or as a separate blood draw. 3) Another way to donate is by requesting that your previous biopsy sample that is stored elsewhere be transferred to the CMD Tissue Repository. Arrangements can be made directly with that facility. 4) The final method of obtaining tissue and organ samples would be during autopsy. A gift of donation made during autopsy will give scientists the ability to study the disease and work toward the ultimate goal of creating a therapy or drug, leading to a cure.

 Stacy Cossette	1. If you would like to make a donation of blood for the establishment of cell lines, a donation of spare tissue from a surgical procedure, or discuss a donation of autopsy tissue, please contact the Repository Manager, Stacy Cossette, at <a href="mailto:scossette@mcw.edu">scossette@mcw.edu</a> or give your permission for her to contact you.
	2. Review the consent paperwork, schedule a telephone consultation with Stacy Cossette, and then email, mail, or fax the signed forms to 414-955-6411.
	3. Your signed consent form will be faxed to your local hospital.
 Dr. Lawlor	4. Dr. Lawlor will establish contact with your surgeon or pathologist, when necessary. 



## Questions?

For more information about the **CMD International Registry (CMDIR)** please contact: Rachel Alvarez, Associate Director at [\(323\) 250-2399](tel:3232502399) or [rachel.alvarez@cmdir.org](mailto:rachel.alvarez@cmdir.org)

For more information about the **CMD BioBank** please contact: Tara Schmidlen, MS CGC, Genetic Counselor at 856-757-4822 or [tschmidl@coriell.org](mailto:tschmidl@coriell.org)

For more information about the **CMD Tissue Repository** please contact: Stacy Cossette, Repository Manager at [scossette@mcw.edu](mailto:scossette@mcw.edu)

## What organizations support the Registry, BioBank, and Tissue Repository?



## Glossary

**Fibroblast:** a connective tissue cell

**Lymphoblast:** a precursor cell that develops into mature white blood cells

**Buffy Coat:** the portion of a blood sample that contains white blood cells and platelets

**Cell line:** a population of constantly dividing cells

**iPS:** induced pluripotent stem cells, also referred to as “man-made” stem cells. A type of pluripotent stem cell artificially created from a non-pluripotent stem cell (often a fibroblast) by forced expression of specific genes.

**Pluripotent:** a stem cell with the ability to develop into multiple different cell types