

# CMD Tissue Repository

## Accelerating Research for Congenital Muscle Disease

*Congenital Muscular Dystrophy • Congenital Myopathy • Congenital Myasthenic Syndrome*

### A Look At 2016

This year was filled with progress and accomplishment. We attended the Nemaline Myopathy Scientific Conference and Family Social in New York, presenting on our biorepository and assisting with a longitudinal study. We hosted a [webinar](#) featuring the work of Dr. Kristen Nowak who is at the Harry Perkins Institute of Medical Research and The University of Western Australia. We also translated our outreach materials into Spanish, French, and German this year to expand into other cultures and countries. We provided informational flyers to the RYR1 and NM Spain conference planning committees. We also hosted an online Meet & Greet.

Much of our efforts in the last year were in serving our researchers. We offer services to deliver tissue samples to them, recruit donations from particular subtypes, and help connect them with other useful resources. Additionally, we distributed some of our centralized tissue from 11 individual participants to three prominent research labs in the United States.

We increased program participation to 113 enrolled and 59 individual donors that have contributed over 280 tissue samples and cell lines now available for research use. The CMD-TR is also pleased to be listed on the [Rare Diseases Human Biospecimens/Biorepositories \(RD-HuB\)](#) website.

### December '16 Issue

- The Year In Review
- NM Conference
- Centralizing Specimens
- Get Counted
- Supporters

The CMD-TR is accepting tissue donations from most regions of the world where language is not a barrier. Visit our website at [mcw.edu/CMDTR](http://mcw.edu/CMDTR)



Kristen Nowak, PhD



**Families attending the Nemaline Myopathy Conference in White Plains, NY**

White Plains, NY was the location of the Nemaline Myopathy Scientific Conference and Family Social in July of 2016 where 145 members of families, individuals with NM, clinicians, and scientists gathered for a weekend of connecting, learning, and fun. Among the attending guest speakers were Drs. Alan Beggs, Coen Ottenheijm, David Roye, John Bach, Michael Lawlor, and Stacy Cossette.

Ms. Cossette [presented](#) on the Congenital Muscle Disease Tissue Repository (CMD-TR) and briefly discussed our two partnered research resources: Congenital Muscle Disease International Registry (CMDIR) and Congenital Muscle Disease Biobank (CMD Biobank), while Dr. Lawlor's presentation detailed the ongoing NM work being performed in his laboratory, including projects on possible metabolic dysfunction in NM and his continuing trials with anti-myostatin therapy in NM mice.

Ms. Cossette and Dr. Lawlor are available, as their schedules permit, to attend congenital muscle disease conferences and present on topics related to biobanking and congenital muscle disease pathology. We also have printable materials that can be shared with conference planning committees to be made available to conference attendees.

Schedules book-up fast, so if your community would like a member of the CMD-TR team to attend your scientific or family conference, please contact Ms. Cossette at [stacy.cossette@cmdir.org](mailto:stacy.cossette@cmdir.org).



## Conference Tips

It was a tremendous opportunity to help plan and attend the scientific and family conference in 2016. For these families, travel can be a sizeable challenge, and making the most of this special occasion requires thoughtful planning and implementation.

1. Start planning early, even a year before the event, and choose your planning team carefully, being as inclusive as is feasible.
2. Resist the urge to pack everything into one weekend. These families are tired.
3. Consider the variety of ages and interests when planning.
4. Allow time for more breaks than you think are needed. Trust me, more are needed.
5. Hire a videographer to record discussion panels, talks, and fun events.
6. Hire a photographer that can be dedicated to capturing moments, documenting events, and can do both large group and individual family shots.

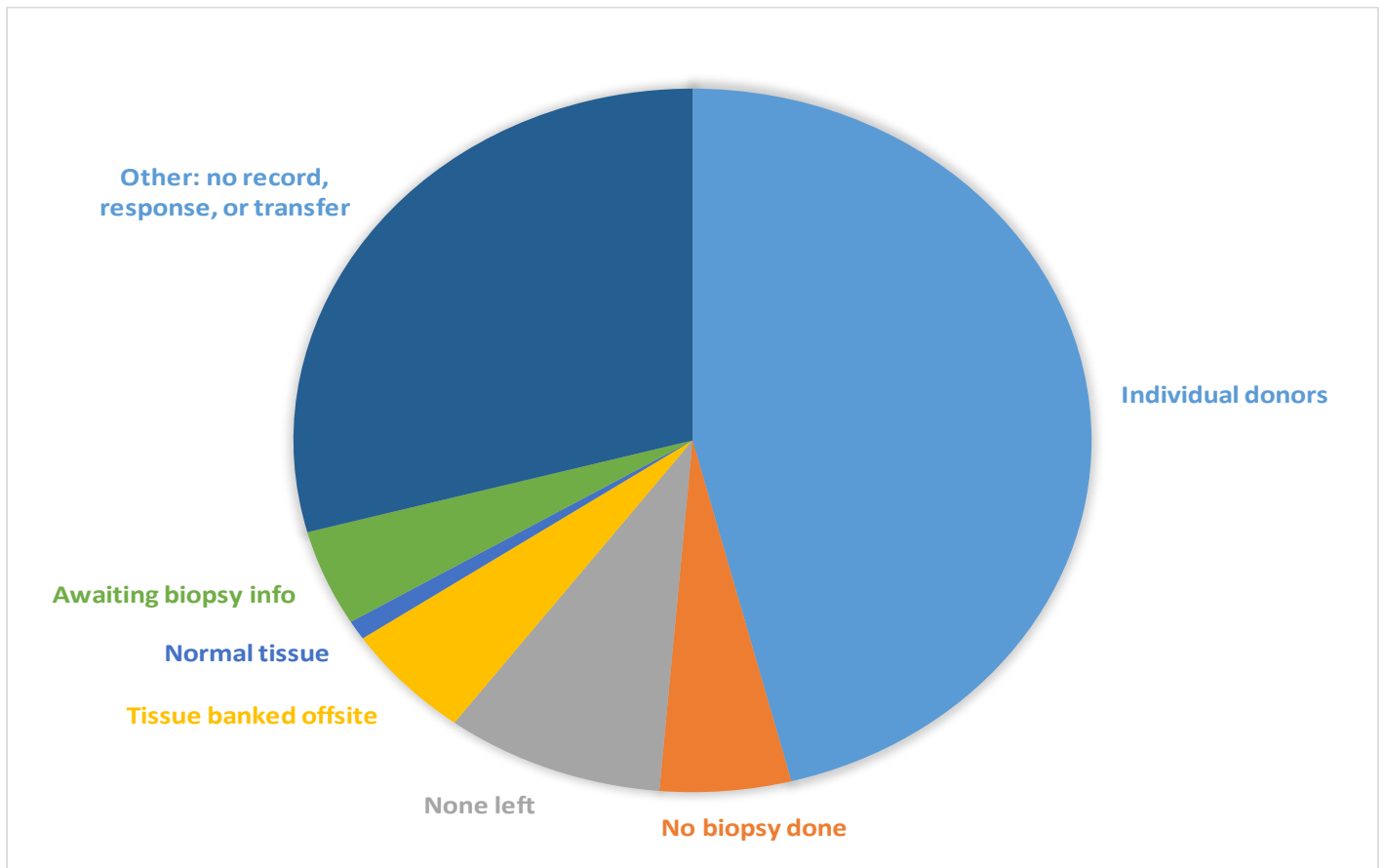


**The all ages event was a great way to meet others affected by nemaline myopathy**



**Clinical research participation was balanced by the day-to-night fun activities**





## The CMD-TR Preserves Your Tissue Specimens

The Congenital Muscle Disease Tissue Repository (CMD-TR) at The Medical College of Wisconsin is a place to store specimens that are donated after diagnostic testing is complete, through routine surgery, or during end-of-life procedures. Centralizing tissue access will make it easier for scientists to obtain appropriate numbers of samples for their research studies.

**Our Mission:** To provide a free service to patients and families interested in supporting medical research and treatment options through tissue centralization.

**Our Purpose:** To make samples from congenital muscle disease (CMD) patients available for research and treatment studies.

**Our Goals:** To establish a large CMD tissue collection and to distribute samples to scientists who study muscle disease.

For a list of all muscle disorder subtypes centralized at the CMD-TR, please visit our partner website, the Congenital Muscle Disease International Registry, at [cmdir.org](http://cmdir.org).

At the CMD-TR, we work in partnership with other research resources to accelerate the pace of research and help individuals and families affected by congenital muscle disease to find the answers and treatments they need.

Throughout 2016, we have been recruiting, organizing data, and assisting in building cell line resources for our researchers. For example, CRISPR-cas9 is a recent technology developed for the targeted editing and regulation of genes that can be applied to congenital muscle disease and uses affected cell lines generated from patient biological sample donations.

For more information on cell lines, contact [stacy.cossette@cmdir.org](mailto:stacy.cossette@cmdir.org) or [tschmidl@coriell.org](mailto:tschmidl@coriell.org)

# GET DONATED.



One of our goals is to create a bank of biological resources that will be used to advance our understanding of congenital muscle diseases. To make this bank a reality, we need you to donate blood and skin samples to the CMD Biobank. Get your free kit today!

Contact Tara Schmidlen:  
[tschmidl@coriell.org](mailto:tschmidl@coriell.org)

Visit for more information:  
[www.curecmd.org/studies/biobank](http://www.curecmd.org/studies/biobank)



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## GET COUNTED!

— connect - register - consent - donate

Please consider making a lasting donation to the CMD Tissue Repository, for yourself, your loved ones, and future generations. The fact that our donors may be able to help people long after they are gone fills them with satisfaction and happiness.

In just a few minutes of your time, you can learn how it works and sign up to be a part of something meaningful.

Contact us today [mcw.edu/cmdtr](http://mcw.edu/cmdtr).

Look for  
announcements  
on our Facebook  
page in  
2017!

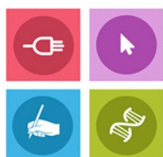


# GET CONSENTED.



If you have had a muscle biopsy, are planning to have a muscle biopsy, or are planning to undergo surgery, consider making a lasting contribution to the CMD Tissue Repository. We need you to consent to have your muscle tissue shared with the CMD Tissue Repository. Contact Stacy Cossette, CMD Tissue Repository Manager, to get started today!

[Stacy.cossette@cmdir.org](mailto:Stacy.cossette@cmdir.org)



## GET COUNTED!

— connect - register - consent - donate



Support for the CMD-TR has been generously provided by:

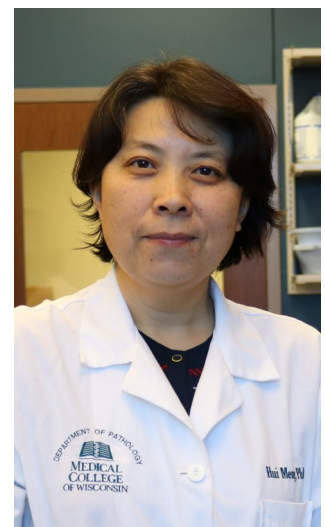
Cure CMD, A Foundation Building Strength for Nemaline Myopathy (AFBS), Where There's a Will There's a Cure, the Joshua Frase Foundation; (JFF), the Rutkowski and Scoggins families, Team Titin, Stacia Creative Photography, the Children's Hospital of Wisconsin Foundation, the Children's Research Institute, and Audentes Therapeutics.



**A FOUNDATION  
BUILDING  
STRENGTH**  
For Nemaline Myopathy



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Dr. Michael Lawlor, Director; Lucinda Lee, Volunteer; Stacy Cossette, Manager; Dr. Hui Meng, Research Scientist