A Look At 2017

It was a year of development and growth. We attended conferences, presenting on our biorepository, meeting families, connecting with researchers and clinicians, and assisting with the mission. We hosted 3 webinars this term: the first featuring the work of Dr. Olga Igoucheva from Thomas Jefferson University in Philadelphia on therapeutic stem cell use in CMD-affected muscles; the second, a discussion group led by Angela Maccaronne, an affected member of the CMD community who is a Master’s degree student in Washington; and finally we had Dr. Reghan Foley of the Neuromuscular and Neurogenetic Disorders of Childhood Section, NIH National Institutes of Health, as a guest speaker to discuss proactive respiratory care in congenital muscle disease (CMD).

As part of our outreach and education, we provided informational flyers at annual conferences and to numerous hospital systems.

Program participation increased to 134 enrolled for tissue donation. We coordinated tissue transfers from 13 medical centers across the United States and Canada. Our donors have contributed over 370 tissue samples in total that are now available for research use. This year we also assisted in the donation efforts of blood and/or skin samples from 60 new families for cell line development.
Attendees of the Cure CMD SciFam Conference in Washington, DC

Washington, DC was the location of the Cure CMD conference in July where 380 members of families, individuals with CMD, clinicians, and scientists gathered for a weekend of connecting, learning, and planning for the future. Among the attending guest speakers was Stacy Cossette, manager of the CMD-TR.

Dr. Michael Lawlor, the CMD-TR Director, has given presentations with Audentes Therapeutics, Inc., Solid Biosciences, and at the MTM family conference, plus several scientific talks at academic institutions, where information on our biorepository was presented.

Ms. Cossette and Dr. Lawlor are available, as their schedules permit, to attend congenital muscle disease conferences or participate via web platform 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.
There are samples stored at the CMD-TR from all across the United States, Canada, Australia, Germany, England, and Romania. All samples are publicly cataloged and made available to researchers anywhere in the world conducting approved studies of the subtypes available.
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, which can accelerate results.

There are many potential research uses for preserved tissue, and in this issue, we will review some of those uses. Genetic studies allow for evaluation of gene and protein expression in the context of a person’s disease, which may identify additional modifiers of disease severity or treatment response.

Single fiber functional studies require tissue samples. Some laboratories can study the process of contraction in frozen tissue samples, allowing for the study of weakness mechanisms in human muscle disease.

Tissue samples are useful for the review and evaluation of pathological findings in these rare diseases. This is a big deal for clinical trials, since having a clear understanding of how disease severity varies from one person to another is important.

Scientists are able to identify which disease models most accurately reflect the muscle diseases of interest through comparisons of findings and markers expressed in each animal model and comparing those to human samples. This applies to cell culture models as well as animal models, since it is important to understand whether the abnormalities seen in these models are actually relevant to the human muscle disease state.

Understanding how biopsy findings or protein expression might predict disease progression or treatment responsiveness is another potential use of specimens. A muscle disease subtype may be highly variable when people are grouped based on their genetic mutations, but there may be certain markers found through tissue sample studies that can more easily predict clinical symptoms and treatment outcomes.

For a list of all muscle disorder subtypes centralized at the CMD-TR, please visit our website mcw.edu/cmdtr or our partner website, the Congenital Muscle Disease International Registry, at cmdir.org.

X-linked myotubular myopathy (XLMTM) is a severe congenital myopathy that has been a focus of our tissue collections. The CMD-TR has assisted in clinical and therapeutic studies in XLMTM, including the INCEPTUS natural history study and ASPIRO investigational gene therapy study that are being performed by Audentes Therapeutics. The gene therapy has shown promising results in animal models, and trials began testing the therapy in affected children in 2017.
Where I Am From

I am from the chains of doubt that bind me to the Earth
and keep me from flying.

I am from the disappointment of dreaming about winning
an Olympic gold medal in a 500-meter sprint,
then waking up seeing my wheelchair.

I am from the sadness that is masked by all of the smiles and laughs.

I am from the heartbreak from watching one of my friends or family walk or run or jump.

I am from the sadness that is masked by all of the smiles and laughs.

I am from the sensation of jealousy when I see a young child giggling and laughing
as they are chased by their older sibling or their parent.

I am from the feeling of being forgotten and isolated from the world.

But, I am also from the adrenaline that courses through my veins,
and racing the winds in my power chair.

I am from the movies and books and stories that provide me with laughter, hope, and satisfaction
when the protagonist jumps over the obstacle they have been facing.

I am from my middle school and wonderful teachers
that provide me with something stronger than steel - knowledge.

I am from the pride that pounds in my heart every time my brother is in a play in football.

I am from my friends, the people who help me stand a little taller,
laugh a little louder, smile a little prouder, and use my imagination freely.

I am from my adrenaline, wheelchair, books, movies, stories, teachers, pride, family and friends.

I am from my Home.

By Kierra Sunris
**In the Know:** If you or your loved one is in a research study or clinical trial, it is important to report changes in health to the study team immediately. This is especially important if you/they get sick, have to go to the hospital, or pass away. Call the contact person for your study site anytime, day or night, to report changes. Make sure to leave a detailed message if you cannot reach someone.

In the tragic case when a person passes away while s/he is part of study, it’s important that a detailed test, called an autopsy, is done to improve our understanding of the disease process and what happened. The capacity of coroners, funeral home staff, or medical examiners to do these sample collections and studies according to protocol is very limited, so please contact the research coordinators about a referral to an academic medical research institution, instead.

It’s important to follow these policies because changes from approved procedure may compromise the validity of the study and the ability to obtain FDA approval to use the treatment, so you are urged to take the advice your study site team offers.

**Looking Forward:** In 2018, we will continue the CMD-TR Invited Speaker Series and plan to have guests from the pharmaceutical industry, a genetic counselor, and more from other specialties and interest areas to inform our communities. Please look for our announcements and messages in the coming weeks and months.
Support for the CMD-TR has been generously provided by:

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Dr. Michael Lawlor, Director; Stacy Cossette, Manager; and the Lawlor Laboratory Science Team