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# CURE CMD

Congenital Muscular Dystrophy

WINTER 2010 EDITION

Together with you, funding our way to a treatment and cure for the CMDs

**Grants funded  
through Cure  
CMD and  
S.A.M.  
partnership:**

**Dr. Denis  
Guttridge, PhD:  
\$50,000 a year  
for two years,  
Ohio State  
University  
Medical Center,  
Columbus,  
Ohio**

**Dr. Dean  
Burkin, PhD:  
\$35,000 a year  
for one year,  
University of  
Nevada, Reno**



As Cure CMD's new chairman, I welcome you to this year's quarterly Cure CMD Newsletter.

We have set an aggressive agenda for 2010 focused on 2 definable goals:

1. register the global CMD population in the CMD International Registry (CMDIR)
2. identify and financially support lead compounds that hold promise as a CMD treatment

If you or your family member has CMD and you have not had the opportunity to register on the CMD International Registry (CMDIR), we encourage you to register within the next month, [www.cmdir.org](http://www.cmdir.org)

**Nevada**

**Dr. Sweta  
Girgenrath,  
PhD: \$50,000 a  
year for two  
years, Boston  
University,  
Boston,  
Massachusetts**

**Dr. Ana  
Ferreiro, MD,  
PhD: \$28,500 a  
year for one  
year, Institute  
of Myology,  
Paris France**

**Dr. Jim Collins,  
MD, PhD:  
\$50,000 a year  
for one year,  
Cincinnati  
Children's  
Hospital  
Medical Center,  
Cincinnati Ohio**

**Also: Merosin  
Deficient CMD  
(MDC1A)  
Clinical Study  
enrolling (see  
below)**



**[www.curecmd.org](http://www.curecmd.org)**

The CMDIR is for all people with CMD. If you do not have a genetic diagnosis, we encourage you to register and indicate CMD, undiagnosed. To achieve momentum, engage the pharmaceutical industry and achieve our mission, we need to reach a critical mass of registered individuals. Your registration is vital to our success.

We have added an additional gene to the CMDIR, RYR1 (ryanodine receptor). People with RYR1 genetic mutations appear similar to SEPN1 CMD.

To take a lead treatment successfully to clinical trial and beyond requires additional work and focus on CMD clinical trial readiness. Cure CMD has co-organized a CMD Outcome Measures meeting in March, 2010 in Amsterdam. The meeting will focus on delivering defined CMD outcome measures. Outcome measures are tools to measure changes in muscle strength, breathing strength and quality of life. In June 2010, we will ask several members of the CMD community to participate in a validation effort to help doctors determine if the measures chosen are valid and apply to people with CMD.

We look forward to seeing you at this year's CMD Family conference (August 14 and 15, 2010).

Warm regards,

Anne Rutkowski MD  
Cure CMD Chairman



**Pennsylvania Teen-Ager with CMD Plans to Compete in Long Branch Half Marathon, Raise Money to Help Find Cure**

For 17-year-old Alexander Van Hook, the Long Branch (NJ) Half Marathon is a challenge he simply can't let slip by.

"Since the Half Marathon will be 13 miles long, it will be quite a challenge for me," he says. "I also love speed blowing in my face and the environment whirring past me. I especially love to race, so I decided to join the Long Branch Half Marathon."

Alex also has another reason to enter the race. The Pennsylvania teen-ager has an undiagnosed sub-type of Congenital Muscular Dystrophy (CMD), a group of diseases that cause muscle weakness at birth or within the first two years of childhood. As a result, Alex gets around in an electric power wheelchair, and that's what he will be using when he joins hundreds of other competitors in the annual event, which

takes place May 2, 2010. The Half Marathon takes place that day along with the 26-mile full marathon.

Alex will be the first-ever power wheelchair user in the race. He will be participating to raise money for Cure CMD, a non-profit organization founded in 2008 that seeks to bring research, treatments and, in the future, a cure for CMD by working globally with parents, researchers, scientists, physicians and government health-related organizations. Cure CMD aims to find and fund research and clinical trials. Success will be determined by clinical applications that improve the lives of those afflicted with CMD.

Cecilia Kodoma has organized Team Cure CMD to roll, walk and run the NJ ½ Marathon in support of CMD awareness and fundraising. It is not too late to join the team or contribute to a team member's \$500 fundraising target. Go to [www.curecmd.org/events/njmarathon2009](http://www.curecmd.org/events/njmarathon2009)

## **CMDIR Update**

**Ahmi Mehta**



The Congenital Muscular Dystrophy International Registry (CMDIR) was officially launched on September 1, 2009 and to date we have 130 participants registered. Thank you to those who have registered and filled out their profiles on the site!

The CMDIR was created to identify the global CMD population for the purpose of raising awareness, creating standards of care, facilitating enrollment in future clinical trials and treatment programs. A patient registry is an extremely valuable tool for rare diseases like CMD. With the future prospect of clinical trials in CMD, The registry is important in identifying which participants could qualify and in disseminating information about those trials to participants. A recent article in the NY Times highlighted the increasing use and value of patient registries (<http://www.nytimes.com/2009/12/22/health/22cyst.html>).

The CMDIR is a comprehensive program that helps participants and providers

- facilitate appropriate diagnosis of CMD
- understand genetic testing in CMD, current options for enhanced testing, and the implications of various genetic mutations found in CMD
- access resources and services to improve care and treatment of symptoms.

One of the goals of the CMDIR is to help participants achieve genetic and/or muscle/skin biopsy confirmation of disease by reviewing existing genetic testing reports and, if necessary, making recommendations for further testing. If genetic testing or muscle/skin biopsy have not been performed, then the CMDIR can help make recommendations on what type of testing might be indicated and/or help participants identify providers experienced with CMD who can help make a diagnosis.

Our goal is to have 500 participants registered on the CMDIR by December 2010. Please help us achieve that goal!

Ami Mehta is the genetic counselor for the CMDIR. In addition to her role as the CMDIR's genetic counselor, she is a pediatric and prenatal genetic counselor at Emory University's Department of Human Genetics in Atlanta, GA.

## **Merosin Deficient (MDC1A) Clinical Biomarker Study**

Cure CMD would like to inform you of an important upcoming Merosin Deficient CMD clinical study with a start date of Feb 1<sup>st</sup>, 2010.

Dr. Jim Collins at Cincinnati Children's Hospital Medical Center (CCHMC) will launch a 1 year trial, Jan/2010- Dec/2010, to identify biomarkers in Merosin Deficient CMD (MDC1A). Dr. Collins and Dr. Carsten Bonnemann at the Children's Hospital of Philadelphia (CHOP) will enroll infants and children with Merosin Deficient CMD between the ages of 0-10 years.

### **Whom do I contact to obtain information about this study with no obligation to participate?**

Shengyong Hu or Paula Morehart (Division of Neurology, Cincinnati Children's Hospital Medical Center)

Phone: 513 636 3202 or 513 636 7451

Email: [shengyong.hu@cchmc.org](mailto:shengyong.hu@cchmc.org)

### **What is the purpose of the study?**

The goal of the study is to find a marker of disease (biomarker) in the blood or urine.

### **Why is it important to have a biomarker specific to MDC1A?**

A biomarker is a critical tool to make clinical trials in MDC1A a reality. A biomarker will allow a clinical trial expert to determine if a drug is having a positive effect on improving disease.

### **Pedal 4 Pierce:**

2009 was a busy year for the Burns family down under. We organized 2 successful fundraisers, 'Help Cure Pierce' and 'Pedal 4 Pierce' bringing in over \$AUS 10,000 for Cure CMD. Traveling to Philadelphia in August 2009 for the CMD Family Conference empowered our family and helped focus our efforts to raise awareness and funds to find a cure.

'Pedal 4 Pierce' was the brain child of 10 year old Christopher Jefferies (a family friend), who decided over dinner one night that he wanted to do something to help our son Pierce and other children like him. From there 'Pedal 4 Pierce' was born with a date set for 12/12/2009. The concept: a family fun bike ride at the picturesque Mt Stromlo Forest Park in Canberra, Australia. Over the next couple of months we started an advertising campaign that spanned the local media; including radio stations, TV stations, newspapers, local magazines and electronic notice boards. People were encouraged to come out to Mt Stromlo for the day with their families and their mountain bikes. Participants were able to donate money either via the Cure CMD website, sponsorship, by buying raffle tickets or donating money on event day!



'Pedal 4 Pierce' was a wonderful day - the weather was perfect and approximately 200 people attended. The event was kicked off with a ride on Pierce's track, a 4km track around Stromlo. Apart from bike riding, we had a cake stall, great raffle prizes and a fabulous sausage sizzle donated by City Select Meats. The event was supported by the Australian Capital Territory MD Association, 5th Gear Racing, a number of the local ONYA Bike stores from around Canberra and various other local businesses.

In 2010, we look forward to another Canberra based fundraiser called 'Putt 4 Pierce' in March/April, a golf fundraiser that will involve local real estate agents. You can find us at [www.helpcurepierce.com](http://www.helpcurepierce.com). The Burns family looks forward to moving to Texas in May 2011 for several years.

### **Update on CMD Legislative Advocacy**

**Jackie Nelson**

*"A small group of thoughtful people could change the world. Indeed, it's the only thing that ever has." - Margaret Mead*

This quote has always inspired me. Dedicated people who come together around shared goals can make important changes that improve lives. We, too - each one of us - can make a direct difference by working together on advocacy initiatives at Cure CMD.



### **We are already making progress.**

In 2009, Cure CMD Outreach and Advocacy focused on meeting key officials to build better relationships with the National Institutes of Health, the Muscular Dystrophy Association and other rare disease groups. We also met with several legislative staffers to introduce our organization and thank them for their past support as well as encourage their continued support on upcoming bills that will affect children and adults with congenital

muscular dystrophy.

In the coming year, we will be calling on parents, family members and friends to join with us in our advocacy work. Our goal is to develop a database of advocates who are willing to write, call or visit their legislators on key issues that affect our children and families. Won't you join us?

It's easy and we'll help you every step of the way. Right now, we are gathering names and email addresses. To sign up, please email me and join our network of advocates at [Jackie.nelson@curecmd.com](mailto:Jackie.nelson@curecmd.com)

Become part of a small group of thoughtful people at Cure CMD and help us make life better for children and adults.

Jackie Nelson  
Director, Outreach and Advocacy  
Cure CMD

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**National Task Force for Childhood Neuromuscular Diseases**

**Attention parents of children with congenital muscular dystrophy (CMD)**

The National Task Force for the Early Identification of Childhood Neuromuscular Disorders is conducting a survey. We want to learn more about parents' first concerns about their child and how they described those concerns to their healthcare providers. The Task Force understands that there may be a long time between when a parent first has worries about an infant or child and when the diagnosis happens. This survey is one part of an outreach program designed to reduce the time to it takes for families to get an accurate diagnosis of neuromuscular disorders.

The Task Force is made up of medical professionals and advocates from several neuromuscular organizations, including CureCMD.

If you have a child diagnosed with **CMD, DMD or SMA**, and if your child was diagnosed between the ages of **6 months and 9 years of age**, you are invited to take part in this brief survey. The survey is completely anonymous and we do not collect any personally identifiable information.

Click here to go to the survey: <https://www.surveymonkey.com/s/97KP7XZ>

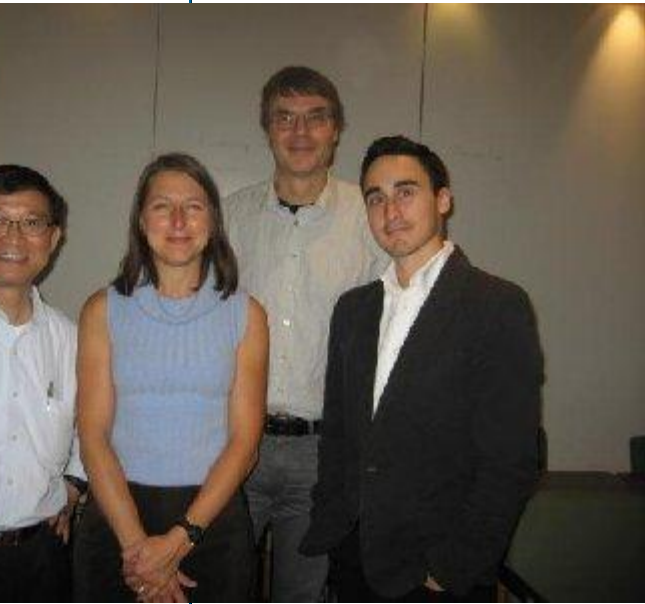
**The deadline for completing the survey is March 15, 2010.**

Thank you for helping us with this project.

If you have any questions about the survey, please contact Jill Hunter at [jill.hunter@curecmd.com](mailto:jill.hunter@curecmd.com)

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**CMD Standard of Care conference a Success**



led by Dr. Ching Wang (US pediatric neurologist), Dr. Thomas Sejersen (neurologist) and Dr. Anne Rutkowski (Cure CMD Chairman)

Dr. Ching Wang, Dr. Anne Rutkowski, Dr. Thomas Sejersen with assistant, Jonathan



**Pictured:**

Back row: Dr. Ronni Cohn, Patricia Jouinot, Dr. Albert Fujak, Vanessa Battista, Dr. Bonnemann, Dr. Andrea Vianello, Dr. Enrico Bertini, Dr. Wolfgang Mueller-Felber, Dr. Muntoni, Dr. Christophe Beroud, Dr. Thomas Sejersen, Dr. Ulrike Schara, Dr. Bob Dr. Ana FERREIRO

Middle row: Dr. Susana Quijano-Roy, Dr. Kate Bushby, Dr. Pam Schuler, Dr. Paolo Aloysius, Dr. Kari Storhaug, Dr. Susan Iannaccone, Dr. Brigitte Estournet, Michelle

Front row: Dr. Ching Wang, Dr. Anne Connolly, Dr. Julaine Florence, Dr. Anne Rutkowski, Dr. Nathalie G

Dr. Vajsar, Dr. Jes Ra  
Dr. Nicolas Deconinck,  
Dr. Reinhard Zel

Dr. Melancini, Dr. Carolin  
Dr. Eagle, Dr. Isabelle Des  
Dr. Rutkowski, Dr. Nathalie G

The international conference which took place on Nov 14th-16th was a great success with concrete plans to move towards publication. A first draft has been executed. The conference will result in the publication of 3 documents:

- a. CMD diagnostic review
- b. CMD care guideline, professional
- c. CMD care guideline, family friendly version

These documents will be written for both the doctor who has only 1 CMD patient in their entire career and the doctor who has several CMD patients. A care guideline will also be written for people with CMD and families. The goal is to provide people with CMD with the tools to seek improved medical care. We believe improved and consistent medical care will impact current "treatment options" for people with CMD.

There are several areas which could not be addressed specifically given the lack of evidence in the literature and varying practice patterns. The goal is to revise the document in 2-5 years with additional information gleaned from studies in CMD patients and a CMD natural history database (to be launched).

The CMD SOC brought together 39 international doctors with expertise in the following areas: palliative care, pulmonary (breathing issues), neurology (seizures, behavior issues, developmental delay), cardiology (heart issues), gastrointestinal (feeding, need for interventions), orthopedics (contractures, scoliosis) and diagnostics. The conference was generously sponsored by: TREAT-NMD, AFM, Cure CMD, Fondazione Telethon and Stanford Medical School.

### **Cure CMD Cookbook**

WANTED: Recipes!!

We are creating a Cure CMD Cookbook! You can help with this Cure CMD fundraiser simply by sending in your favorite recipes.

Everyone is encouraged to submit recipes--feel free to ask friends, co-workers, acquaintances, church members, etc. to submit their recipes. There is no obligation to purchase a cookbook if you submit a recipe. Your first and last name will be displayed next to each recipe you submit.

The cookbooks will be completed in spring of 2010 and will be available for the general public to purchase. Recipes do NOT have to be created by the sender--they can be recipes handed down from families, or favorite ones from cookbooks, the internet, etc. Just please make sure they have been tried and are yummy!

Please email your recipes to [anncloudcurecmd@gmail.com](mailto:anncloudcurecmd@gmail.com) no later than February 15, 2010. Each person is limited to 5 recipes. All food categories are welcome! Thank you!!

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### **CALENDAR OF EVENTS 2010**

1. CMD ENMC Outcome Measures workshop, March 5th-7th, 2010, Naarden, Holland
2. S.A.M. Emerald Ball, March 13<sup>th</sup>, 2010, Hotel Europa, Belfast
3. Aidan Gowarty CMD Fundraiser, Lehigh Valley Iron Pigs Baseball Stadium, Allentown, Pennsylvania, April 21st, 2010
4. Team Cure CMD runs, walks, rolls the NJ ½ Marathon, May 2<sup>nd</sup>, 2010, Long Branch, NJ
5. Ottawa Conference on New Directions in Biology and Disease of Skeletal Muscle, May 5-8th, 2010 Ottawa, Ontario, Canada, <http://www.med.upenn.edu/muscle/registration.shtml>
6. Project CMD Outcome Measure Validation, June 12<sup>th</sup>-13th, NIH, DC
7. Kyra's Crusaders Family Fundraiser, June 26, 2010
8. CMD Family Conference, Summer 2010 August 14-15, 2010
9. CMD Translational Grant Award Deadline, Sept 15th, 2010
10. [\*\*World Muscle Society, Oct 12th-16th, 2010, Kumamoto, Japan\*\*](#)
11. Team Cure CMD runs, walks the San Francisco ½ Marathon, Nov 7<sup>th</sup>, 2010