



# The PMD Foundation

## Newsletter

Striving for a World Without PMD

Vol. IX Issue 1  
Spring 2014

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**WE HAVE MOVED: Please note our new corporate address:**  
 PMD Foundation  
 1 Green Tree Center  
 10000 Lincoln Drive East, Suite 201  
 Marlton, NJ 08053

## Chad Chlebowski — Fairfield, Connecticut



Chad Chlebowski is 5 years old and lives in Fairfield, Connecticut with his sister Jillian, twin brother Ian, and parents Angela and Jon. He is such a happy boy, friends always say that he lights up a room. The Chlebowski family is always joking, and although Chad speaks quietly, his mother suggests we listen carefully, because he is most likely telling a joke. He is always the first one to crack a joke or give a hardy laugh. Chad's good nature really makes him an inspiration to all who meet him.

Chad had a difficult infancy, his family was unsure what was wrong. Like many PMD children, he was initially misdiagnosed with a variety of diseases. After his fifth birthday, following testing at Johns Hopkins Hospital in Baltimore, he

was diagnosed with PMD null syndrome.

Chad can walk with a walker, but he tires easily, so a wheelchair is needed for long distances or at the end of the day. He does need assistance with many daily living activities. Chad is in mainstream school where he has many friends.

Chad's favorite activities include playing soccer and video games. He will be starting baseball this spring in a Challenger League and his parents are sure he will love it. His favorite foods are those of most kids.....treats....chocolate....and French toast!



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## Editor's Message

Welcome to the Spring issue, although it sure doesn't feel like it. It is still in the 30's in NYC and there are still shrinking piles of dirty snow in the corners. But I hope that everyone had a great Holiday season before what was a pretty brutal winter. I know that many of you have had the pleasure of trying to push a wheelchair through the snow, and for those of you in Florida or a similar climate; it is an experience that you don't want to try, even once.

The issue is earlier than usual because the Golf Outing was

pushed up to May 14<sup>th</sup>. The theme for the 13<sup>th</sup> Annual Outing is Oktoberfest. Linda Leonard always guarantees that it will be a great event with her enthusiasm. She and her team of volunteers seem to surpass everyone's expectations on an annual basis. Try to come out and support her and the Foundation this year.

Another upcoming event of note is the 25<sup>th</sup> Anniversary Family Conference. It will take place from June 12-15 in Indianapolis. Come see old friends and meet new ones as we celebrate our

families with Mama Bear Patti Daviau.

The latest update on the PMD Registry is that we have over 110 contact addresses in the database. We do appreciate everyone that responded to our previous requests and have contributed to the first stage in creating a medically viable registry. Even though we have taken the initial steps, a true registry must contain a variety of data. Now, keep in mind that in order for researchers to even consider a project for a rare

*(Continued on page 4)*

**NEWSLETTER**  
**Rob Gasperetti**  
 Editor  
**Kathy Kanous & Don Hobson**  
 Publishers

# Research Updates

## THE PMD PATIENT REGISTRY

**There was an important job to be done and Everybody was asked to do it.  
Everybody was sure Somebody would do it.  
Anybody could have done it, but Nobody did it.  
Somebody got angry about that because it was Everybody's job.  
Everybody thought Anybody could do it, but Nobody realized that Everybody wouldn't do it.**

Sound confusing? When you sort it all out, it is a commentary on human nature. Many people have good intentions, but things happen that get in the way, so tasks are left to others because "surely they have more time." Unfortunately, families with PMD-affected children (and adults) all lead busier than normal lives. We know that time is precious when everyday is filled with therapies, appointments and other care-giving tasks.

However, we now face the fact that almost no one has taken the time to submit their information to the PMD Registry. Much effort has gone into creating the collaboration with Dr. Adeline Vanderver at Children's National in Washington DC. They have been ready to start collecting the data for months now, but only five families have contacted them and only **ONE FAMILY** has followed up. The PMD Foundation has emphasized the value (actually the need) to create a PMD Registry if progress is to be made in PMD research. The opportunity now exists, but it is up to you, the PMD families, to bring this to fruition. Please take the time to make contact and contribute your family's information. For more information click [here](#).

For those of you who have filled out countless forms in the past, there may be a shortcut. If you contact the doctor or researcher who currently holds that information and ask them to send it to you (after all, it is yours), you could then use it to fill out the secure online questionnaire without having to recollect everything. Even if the older form doesn't contain all the data now being requested, it's a start.

The PMD Foundation has also been in contact with STEM CELLS Inc. regarding their future plans for clinical trials. It is obvious that a certain level of patient data will be required to choose future candidates. If that data is readily available, it will serve to expedite the selection process. Yet, this is only one of a number of PMD research programs on the horizon. Even if you feel your child will not benefit from these prospective medical advances, submitting your data may help both younger and future patients as part of a larger body of PMD medical history. Please take the time to become a part of this very important project.

**If you have questions, call Amy Pizzino at Children's National, (202) 476-4975. We are also hoping to have Amy attend the Riley PMD Family Conference in Indianapolis on Friday so you can ask her questions face-to-face**

## Participants Still Needed for WSU Studies

The Neurogenetics group at Wayne State University is carrying out two studies to analyze patients with Pelizaeus-Merzbacher Disease (PMD). These studies are described below.

The first study, funded by the European Leukodystrophy Association, is to obtain clinical and MRI data from individuals with PMD who have a PLP1 gene duplication. The purpose of this study is to characterize the changes in the white matter or myelinated regions of the brain in order to better understand how the gene duplication produces clinical symptoms and signs in these patients. This is important, since future treatments for this form of PMD depend on reversing or modifying these changes. Eligible patient must have a PLP1 duplication and should be at least 9 years old. Study patients will travel to our medical center in Detroit, MI where they will undergo an MRI scan and a detailed neurological examination. No anesthesia or sedation will be used in this study, and no invasive procedures will be performed. Patients and their family will be reimbursed for both travel costs to Detroit and their housing while there.

The second study is a "natural history" study of patients with PMD. The purpose of this study is to track the clinical history and progression of individuals with PMD caused by any type of mutation in PLP1, including gene duplications. This study is important, since it will provide the background data critical for assessing future treatments for PMD. The efficacy of drugs and/or other interventions in patients with PMD must be judged against the known natural history of the disease, and these data can only be obtained from PMD patients followed over time. All patients with PLP1 mutations are eligible for this study. The study requires filling out several forms and a telephone interview. No travel is involved in this study, and there will be no monetary reimbursement.

If you would like to participate in either of these studies, please contact either Dr. John Kamholz, and/or Ms. Jas Sohi at one of the numbers listed below.

John Kamholz MD, PhD  
Professor of Neurology and Molecular Medicine and Genetics  
Email: [jkamholz@med.wayne.edu](mailto:jkamholz@med.wayne.edu)  
Phone: 1-313-577-0925

Ms. Jas Sohi  
Study Coordinator  
Email: [aa1246@wayne.edu](mailto:aa1246@wayne.edu)  
Phone: 1-313-577-8610



Thank you in advance for your willingness to participate in these important studies.

# Research Updates – continued

## Introducing Dr. Jeremy Laukka



We are pleased to introduce Jeremy Laukka, Ph.D., as the newest member of the PMD Foundation Scientific Advisory Committee. Dr. Laukka is currently an Assistant Professor of Neuroscience and Neurology at the University Of Toledo College Of Medicine. As such, he spends six months of the year teaching medical students and residents, and the remainder in scholarship work. He spends all of his research energy on Pelizaeus-Merzbacher Disease.

Dr. Laukka received his B.S. from Michigan State University and his M.S. and Ph.D. from Wayne State University School of Medicine in Detroit. While there, his advisor was the late Dr. Jim Garbern. He also did extensive work with the current acting Chairman of the Scientific Advisory Committee, Dr. John Kamholz. He is working on several research articles on PMD. The most recent was as the first author of an article published in the *Journal of Neurological Sciences* that was mentioned in the December issue of the PMD Foundation newsletter. That was the culmination of a project that took six years. As a member of the S.A.C., he and the other members of the committee are responsible for reviewing and scoring research proposals that are seeking funding from the Foundation. He is the first addition to the S.A.C. since its founding.

Dr. Laukka resides in Michigan with his wife, Nicole, and his son, Dallin. Along with being an avid Chicago Bulls fan, he enjoys travel, water sports, and sailing in his limited amount of free time. We welcome him to our fight to eradicate PMD and thank him for his service to date in that mission.

## Just For Fun

Looking for a spring time art project you can share with your child? Check out **Kinder Art** by clicking [here](#).



"It's spring fever. That is what the name of it is. And when you've got it, you want - oh, you don't quite know what it is you do want, but it just fairly makes your heart ache, you want it so!"  
~ Mark Twain



The first day of Spring is one thing, and the first Spring day is another. The difference between them is sometimes as great as a month.  
Henry Van Dyke



## Editor's Message (continued)

(Continued from page 1)

disease such as Pelizaeus-Merzbacher, they would need to know how many patients are available, where they are, their family history, and the natural history of their disease. Please read the article on page 2 to discover how you can take the next step, by contacting Children's National Hospital in Washington, D.C. Even if you did not initially supply your contact information, you are still encouraged to reach out

and join our effort.

It is my pleasure to welcome Dr. Jeremy Laukka to the PMD Foundation Scientific Advisory Committee. He currently is an Assistant Professor of Neurosciences and Neurology at the University of Toledo. He was previously at Wayne State University where he worked with Dr. Jim Garbern and Dr. John Kamholz. Neither of those names are unfamiliar to the PMD community. Dr. Laukka also was a co-author of the published article from the December newsletter.

You can find that, and all past newsletters, in the website archives [here](#). We thank Dr. Laukka for his past research into finding a treatment and cure for PMD and welcome him to the fight as a member of the S.A.C.

That's all for now. I hope to see many of you at the golf outing and family conference.

Rob Gasperetti

## More From the Editor

In most issues, I ask people to submit articles about their lives with their PMD kids or ways that they make their lives easier. Unfortunately, I rarely get a submission. That is why the *Newsletter* only comes out three times per year. There is only so much to write about. So please try to get me something (anything) for future issues. The next one is due in July and will focus on the Family Meeting and the Golf Outing so there will be room to hear more about YOU!

So, because I still have some room for this issue, I will leave you with a story about our ski trip with NEDS. Hope you enjoy it, but I want to read more about your family next time.

To submit something, just email it to [treasurer@pmdfoundation.org](mailto:treasurer@pmdfoundation.org) or just message me on Facebook. Thanks.

Rob Gasperetti

## NEDS – New England Disabled Sports



Our PMD kids are a blessing, and we know it. However, by having children with disabilities, we need to make sacrifices, especially at vacation time. Camping trips are usually for drive-up sites removing any hope of a wilderness experience, and action sports are often eliminated or reduced to being a spectator sport. What we all have in common is that we just want our PMD family members to feel like they are part of the family activities.

However, there is a chance for the kids to experience the thrills that others have. There are a number of groups that can assist with sports. One such group is New England Disabled Sports (NEDS). Their group of volunteers is willing to assist disabled children and adults with an array of sports, including skiing, cycling, surfing, archery and more. In the photo, you see Brian Gasperetti (17) skiing at Loon Mountain, NH. He had a blast and made it all the way to the summit and back in his ski sled. He and his brother Dylan (10) were accompanied by at least three volunteers at all times. The sleds adapt to ride up the chairlifts, but without the inconvenience of waiting in lift lines. You can visit the NEDS website at [www.nedisabledsports.org](http://www.nedisabledsports.org) to see what other activities they offer. All of

the volunteers were happy to share their time with them, and despite the 10 degree weather, their attitudes warmed our hearts.

N.E.D.S. was founded in 1984 as White Mountain Adaptive Snow Sports and was renamed in 2008. They currently operate out of Loon Mountain and Bretton Woods, both in New Hampshire. They currently have 259 specially trained volunteers. On our recent trip, I was able to see a one legged skier training a volunteer with two legs in how to ski on one leg with outriggers. It was truly a sight to see.

To date for this season, they have provided over 2,700 lessons to over 390 disabled athletes, and I'm sure they all smiled for each of them. Their mission just sums it up; "through sports, to change lives affected by disabilities." I can think of two lives that were changed over one weekend.

For those of you who are not in the Northeast, N.E.D.S. is a chapter of Disabled Sports USA. That is a network of over 100 organizations dedicated to helping the disabled do just what mostly everyone else can. Their website is [www.disabledsportsUSA.org](http://www.disabledsportsUSA.org). There you will find links to local organizations that are more than willing to help. They offer sports that you thought your PMD kids would never get the experience to enjoy. The memories will last forever, for both you and your child (or adult.) And be sure to take pictures and send them in, as I am sure that other families would love to see them and hear your story.



# Upcoming Events

## 13th Annual PMD Golf Outing — May 14, 2014



For all of you who have always wanted to visit Bavaria after the harvest season but couldn't get away, we have the next best option.

Oktoberfest will be recreated by PMD mom Linda Leonard and her volunteer crew of frauleins on Wednesday, May 14 at the Cranbury Golf Club in West Windsor, NJ. The festivities will begin with a barbecue lunch at 11:30 AM followed by a shotgun start at 12:30. Golf will run until 5:30 PM, and then the real fun begins. Live music and cocktails will commence followed by dinner and a wide assortment of raffle prizes and auction items. Non-golfers are always encouraged to come for cocktails and dinner. We are not sure of what Linda has in store, but we are sure it will be a great evening! Cranbury Golf Club has even approved the wearing of lederhosen as appropriate golf attire.

Please try to come down to the Princeton area to support the event. You are supporting a great cause and a great effort and are destined to come home with some great memories. That is even if you slice every shot into the woods. Just be sure to save your best tee shot for the 16<sup>th</sup> hole. Trust me on this.

You get all of this fun for only \$195.00. The cocktails and dinner package is only \$75.00. If you can't make the event but want to support the event with a raffle prize donation, you can contact Linda at [leonard@pmdfoundation.org](mailto:leonard@pmdfoundation.org). Reservations for the golf and dinner package, or the dinner and cocktails only package can be made online at [www.events.org/2014PMDgolfouting](http://www.events.org/2014PMDgolfouting). Bring your friends too! You can also make a cash donation at that site.

We're hoping to see old (and some new) friends at the outing. Auf wiedersehen.

## 25th Annual Riley PMD Family Conference June 12-16, 2014



An annual event that is written in stone on many PMD family's calendars is celebrating its 25<sup>th</sup> Anniversary this year. Of course, it is the Family Conference at the Crowne Plaza – Downtown Union Station, in Indianapolis, Indiana. This year's event will take place from Thursday, June 12<sup>th</sup> through Sunday the 15<sup>th</sup>. Patti Daviau has once again arranged for the services of the medical staff of Riley Children's Hospital to volunteer their talents for the conference. They will be seeing patients at the hospital on Friday and giving presentations at the hotel on Saturday morning. Around those events, there will also be plenty of family gatherings and catching up with old friends.

Attractions in past years have been trips to the Indianapolis Zoo, a stroll at White River State Park, the Children's Museum of Indianapolis, tours of Lucas Oil Stadium (the home of the Indianapolis Colts) or a trip to the ballpark to see the Indianapolis Indians, the AAA affiliate of the Pittsburgh Pirates. Of course, Indianapolis Motor Speedway is just a short drive away. However, most of the memories come from the (very) late chatting sessions in the lobby of the Crowne Plaza in the

company of the ghosts of Union Station, which are present throughout the lobby.

To reserve a room, please call the hotel directly at (317) 631-2221. Mention that you are with the Riley PMD Group. There are a limited number of rooms on reserve at a rate of \$99.00 per night. (The current on-line rate is \$254.00 per night.) There is no cost for conference attendance, just your own travel, entertainment, and meal costs.

Please try to join us as we celebrate Patti's 25<sup>th</sup> Anniversary of running this event. It is always great to see old friends, and of course, it is equally great to meet new ones.

## 6th Annual Jones PMD Walk 'n Roll — September 13, 2014

It's still a few months away, but don't forget to Save the Date!

WALKING for a CAUSE, ROLLING for a CURE -- Raising awareness and funds for the PMD Foundation to fight against Pelizaeus Merzbacher Disease and Support PMD Families

It's a 2.5 mile family fun walk and roll (wheelchairs, wagons, strollers and children's bikes allowed) along Chicago's lakefront, near the Museum of Science and Industry, from 63rd Street Beach to Promontory Point and back. Following the walk, there will be a variety of family friendly activities (including a raffle with great prizes) and refreshments will be served. Keep an eye out for updates with more information to follow.



# Fundraising – Use Amazon Smile to Shop Online



## AMAZON SMILE

There is a way that you can help the PMD Foundation with just five strokes of a keyboard.

Most people use *Amazon.com* when they do their online shopping. Amazon has excelled at making shopping effortless and they have most everything you would need. But did you know that they help charities on a daily basis? All you have to do is log on to <http://smile.amazon.com> and enter the PMD Foundation as your charity of choice. Then use the same address when doing your Amazon shopping and .5% of eligible purchases will be donated to the Foundation. If you forget the *smile* part the next time you log on with that computer, they will remind you and link you to the donation site. It is the same company, same prices, same website.

So the next time you need to do some online shopping, just remember to Smile.

## GOODSEARCH/ GOODSHOP

What, you haven't finished your Holiday shopping yet?!? It may be time to go online to finish it up. When you do, try *Goodshop* from *Goodsearch.com*. Go to [www.goodsearch.com](http://www.goodsearch.com) and enter what you are looking for in the search box. Then go to the stores with the yellow links. Or if you know exactly what you are looking for, go to the *Goodshop* icon and choose your store.

Just make sure that you list PMD Foundation of Franklin Square, NY as your chosen cause. You don't pay anything extra, and even have easy links to coupons for additional discounts on the goods or shipping. The donation to the Foundation is paid by the merchants themselves. Each year, we receive in excess of \$200 simply by your use of the search feature and shopping with Goodshop. It is powered by Yahoo, so you know it is secure.

## GOOD DINING (online shopping)

We truly appreciate everyone who has used GoodSearch as their primary search engine and for stopping by GoodShop when doing all of their online holiday shopping. Every penny helps, and that is a painless way to help the PMD Foundation.

Now the people at Yahoo who brought you those programs have started GoodDining. First, you register your credit cards at [www.goodsearch.com](http://www.goodsearch.com). Then when you dine out at one of their participating restaurants, a donation of up to 6% of your total check will be sent to the PMD Foundation. It won't cost you anything additional. You can even use it to find a restaurant, keep track of your favorites, and rate and review your dining experience. Bon appetit!



## iGIVE (online shopping)

Shopping or searching for purchases through iGIVE can result in a donation for the PMD Foundation. A penny or more per search, a \$5 bonus for your first online purchase and up to 26% of your purchases will be contributed to the PMDF.

Over 700 stores participate in this donation program, including Amazon.com, eBay, Staples, JCPenney, Barnes & Noble, Overstock.com, Office Depot, QVC, Home Depot, HSN, Gap, & NORDSTROM. Plus, as an iGive member, you SAVE money with exclusive coupons and free shipping deals.

It's easy. Select the PMDF as your favorite cause, register with iGive and shop at brand name online stores through the iGive Mall. Logon today to:

[www.iGive.com](http://www.iGive.com)

## DONATION LINE LLC (vehicle donation center)



RUNNING OR NOT  
AND GET A TAX DEDUCTION

Pelizaues-Merzbacher Disease  
(PMD) Foundation

Use Donation Line to donate cars, trucks, motorcycles, RVs, boats, jet skis or snowmobiles to benefit the PMD Foundation and get a tax deduction for yourself. They provide FAST, FREE pick-up of your vehicle (running or not) and have an A+ rating by the Better Business Bureau. The process can be initiated 24 hours/7 days a week by phone at 1-877-227-7487 (ext. 2434 for PMDF) or by filling out their online form. A towing agent will contact you to arrange a pick-up time, and provide you with a pick-up receipt. The vehicle will be sold at auction and the PMD Foundation will send you a tax-deduction letter upon receiving the funds. Get more information about tax deductions for donated vehicles, at:

[www.donationline.com](http://www.donationline.com)

## SUPPORT THE CAUSE — IT'S EASY TO DONATE

The Foundation is a non-profit 501(c)3 organization. Your contributions are tax deductible to the extent allowable by law. ALL participants of The PMD Foundation serve as **VOLUNTEERS**, however, operating costs and research funding are expenses we face to keep the Foundation viable. **We need your financial support**, it's that simple.

Click [here](#) to make your contribution by credit card or Paypal. If sending a check, please use the address on the last page of the newsletter.



### **The PMD Foundation**

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Websites:

[www.pmdfoundation.org](http://www.pmdfoundation.org) &  
[www.facebook.com/ThePmdFoundation](http://www.facebook.com/ThePmdFoundation)

**Together, we can make 2014 the year that a treatment is discovered, leading to an eventual cure!**

**Please make a donation either:**

**By visiting our website to make a safe and secure contribution**

[www.pmdfoundation.org/donation.htm](http://www.pmdfoundation.org/donation.htm)

**Or**

**By clipping the following coupon and mailing it with your check**

### **Our Vision:**

**A World Where There Is No PMD**

### **Our Mission:**

- Provide support for families affected by PMD
- Raise public awareness and support for PMD
- Fund research of Pelizaeus-Merzbacher Disease

## **My Donation**

**I would like to support the PMD Foundation's mission of family support, awareness and research.**

**Enclosed is my tax-deductible donation of \$\_\_\_\_\_.**

I would like these funds designated for \_\_\_\_\_

Name: \_\_\_\_\_

Address: \_\_\_\_\_

City: \_\_\_\_\_ State: \_\_\_\_\_ Zip: \_\_\_\_\_

Please detach and mail with your contribution to our Treasurer's address at:

**The PMD Foundation**

**1059 Mosefan Street**

**Franklin Square, NY 11010**

***THANK YOU FOR YOUR SUPPORT!***