



# Fall 2012 E-Newsletter

## From the President

5147 S. Harvard Ave  
Suite 181  
Tulsa, Oklahoma 74135

PH: 347.470.4373

FAX: 918.516.0227

EM: [contact@thegfpd.org](mailto:contact@thegfpd.org)

WEB: [www.thegfpd.org](http://www.thegfpd.org)

### BOARD MEMBERS

**Shannon Butalla**  
President  
[shannon@thegfpd.org](mailto:shannon@thegfpd.org)  
402.429.5650

**Anne Park Hopkins**  
Vice President  
[annepark@thegfpd.org](mailto:annepark@thegfpd.org)

**Melissa Bryce Gamble**  
Treasurer  
[melissa@thegfpd.org](mailto:melissa@thegfpd.org)  
918.230.7713

**Geoff Edgar**  
Secretary  
[geoff@thegfpd.org](mailto:geoff@thegfpd.org)

**Mousumi Bose**  
Medical/Scientific Liason  
[mousumi@thegfpd.org](mailto:mousumi@thegfpd.org)

**Pamela Marshall**  
Registry & Support Group  
Coordinator  
[registry@thegfpd.org](mailto:registry@thegfpd.org)

**Woody Woodbury**  
[woody@thegfpd.org](mailto:woody@thegfpd.org)

There is much to celebrate as The Global Foundation for Peroxisomal Disorders had its second birthday October 5<sup>th</sup>! The combined efforts of our board members, medical and scientific advisors, families, and friends has helped GFPD grow and evolve to make an impact for children diagnosed with Peroxisome Biogenesis Disorders – Zellweger Spectrum Disorders (PBD-ZSD).

In July we hosted our second annual conference in Orlando, Florida. Present were twenty-eight individuals with PBD-ZSD and nearly 150 family members, friends, and medical professionals passionate about this spectrum of disorders. The 2012 Family Support Conference was an amazing time for families to meet other families sharing common experiences - and feel, for a few days of the year, that they are not alone. Our 2013 Conference will be held at The Downtown Embassy Suites Hotel & Conference Center in Lincoln, Nebraska July 26 – 30<sup>th</sup> and focus on medical management and family support.

The GFPD is assisting Dr. Nancy Braverman (McGill University, Montreal, Quebec, Canada) with her efforts to coordinate a Natural History Study (the first of its kind) to collect longitudinal data on PBD-ZSD. Another exciting development is progress toward a drug trial for patients with PBD-ZSD. The pharmacological agent tested in the lab has shown to improve peroxisome function in skin fibroblasts, which holds promise for those affected.

We owe much appreciation to our outgoing board members, Carolina Alfaro and Tracey Partridge. Their help launching GFPD was invaluable and we appreciate their two years of service. We welcome two new board members- Mousumi Bose, our Research and Medical Liason and Pamela Marshall, our Family Registry and Support Group Coordinator.

Many exciting things are to come for The Global Foundation for Peroxisomal Disorders. Please keep up with us on our website [www.thegfpd.org](http://www.thegfpd.org) and on our public Facebook page at [www.facebook.com/GlobalFoundPD](http://www.facebook.com/GlobalFoundPD).

Shannon Butalla  
President, GFPD



## **RESEARCH NEWS**

### **Natural History Study of Peroxisomal Biogenesis Disorders**

GFPD is assisting Dr. Nancy Braverman, M.S., M.D. (Associate Professor Depts. of Human Genetics and Pediatrics McGill University-Montreal Children's Hospital) with her efforts to recruit patients for a Longitudinal Natural History Study of Peroxisomal Biogenesis Disorders.

For more information, please visit [www.clinicaltrials.gov](http://www.clinicaltrials.gov), a service of the U.S. National Institutes of Health or follow this direct [link](#). If you are a parent/ guardian of a child or adult with a Peroxisomal Biogenesis Disorder and wish to be involved in the study, please print off the [Natural History Study Consent Form](#) and [Photo Release](#) and mail original signed copies to: GFPD- 5147 S. Harvard Ave. Suite 181 - Tulsa, OK 74135.

If you have any questions about the Natural History Study or your involvement in it, please contact Melissa Gamble by email at [melissa@thegfpd.org](mailto:melissa@thegfpd.org). **This study is open to all families in the US and Canada with living and/or deceased children.**

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### **The ALD-AMN Global Alliance and Newborn Screening Project**

While PBD-ZSD and ALD (or X-ALD) are different diseases, they are related peroxisomal disorders that share many similarities. The newborn screening that has been developed to identify boys with X-ALD should also identify children with PBD-ZSD, D-bifunctional protein deficiency (DBPD) and other peroxisomal disorders.

The [ALD-AMN Global Alliance](#) is supporting efforts in the United States and the United Kingdom to make this screening a recommended screening for all newborns and they are currently supporting a study led by Dr. Tortorelli at the Mayo Clinic that is collecting data on the effectiveness of the screening/test. However, more Dried Blood Spots (DBS) are needed.

GFPD is encouraging our families to participate. DBS from individuals who have been diagnosed with PBD-ZSD and DBPD in addition to those of known carriers are needed for the study. For more information please visit the ALD-AMN Global Alliance [website](#) or contact Dr. Tortorelli at Mayo Clinic directly by phone (507-266-8158) or email ([biochemicalgenetics@mayo.edu](mailto:biochemicalgenetics@mayo.edu)).

Although at this time, treatment for PBD-ZSD and DBPD are symptomatic, an early and correct diagnosis can make a difference for families. Many families wait months and even years before receiving a diagnosis of a PBD-ZSD or DBPD, many receiving multiple incorrect diagnoses along the way. If children were identified at birth, families will not have to undergo months and years of searching for the cause of their child's delays and health issues. Families will also be able to work with their medical team to be proactive in the treatment and intervention of hearing loss, vision loss, seizures, bleeding issues, adrenal insufficiency and any other symptoms that arise.

Additionally families who have a child who has been identified to have X- ALD, a PBD-ZSD, or DBPD through a newborn screening will also be aware of their carrier status and thus be able to make an informed decision about the risks and options for future family building.

**This is an international study and we hope that all of our families will choose to participate.**

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## **UPCOMING EVENTS**

### **Art for Archer, Meridian, MS - October 20, 2012**

For more information visit [www.artforarcher.com](http://www.artforarcher.com) and/or contact Ashley Maple: at [ashleynmaple@yahoo.com](mailto:ashleynmaple@yahoo.com)

### **Painting for PBDs, Louisville, KY - October 21, 2012**

For more information visit [www.facebook.com/events/103258656497201/](http://www.facebook.com/events/103258656497201/) and/or contact Bethany Barno at [bbarno@amazon.com](mailto:bbarno@amazon.com)

### **2013 GFPD Conference in Lincoln, NE – July 26-30, 2013**

The 2013 GFPD Conference will be held at the Embassy Suites—Lincoln, in Lincoln, Nebraska from July 26th through the 30th. More information will be available soon.

## Birthdays

### October

2 Nicola Peraro  
 5 Cameron Chambers  
 6 Sarah Brownsworth  
   Clayton Murphy  
 8 William Tairo\*  
 13 Teagyn Smith-Hendricks\*  
 14 Kenna Maag  
   Liam O'Connell  
 16 Parker Owens\*  
 17 Axel Drienuizen  
   Thomas O'Connor  
 19 Gabriela Castaneda\*  
   Ethan Tulloch\*  
 25 Joshua Dilworth\*  
 26 Levi Berg\*  
 29 Savanna Watts  
 30 Kayla Helbert  
   Gonzalo Rodrigo  
   Charleigh Tomes

### November

1 Kyle Woodford\*  
 2 Michael Passerino\*  
 5 Sienna Castro\*  
   Peter Hopkins\*  
 9 Skyler Gratton  
   Ainsley Wade  
 11 Ashley Jess  
 15 Natalie Morford\*  
 16 Jonathan Korsholm  
   Gregory Masten  
   Justin Pires\*  
 20 Phoenix Smith  
 21 Vincent Carusone\*  
   Millie McMenemy  
 23 Hope Guthrie\*  
 29 Alex Callahan\*  
 30 Nicholas Braga\*  
   Elisa Brechet\*

### December

3 Justin Comeau  
   Erik Gouker\*  
   Matthew Wulf  
 4 Lexie Harrison  
 7 Benjamin Loustau\*  
 12 Paulina Adrino\*  
 13 Thomas Wallace\*  
 14 Josh Boyd  
   Logan Diderrich\*  
 19 Corrine Massey  
 21 Makenzie Leonhardt\*  
 23 Ciera Charbonneau  
 24 Lilly Dickson\*  
 25 Alexander Clark\*  
 28 David Wooten\*  
 29 Jaiden Dewsnap\*  
 30 Mohammed Domi\*  
   Marie Lawson  
 31 Alyssa Patrick\*



\*deceased

## Anniversaries

### October

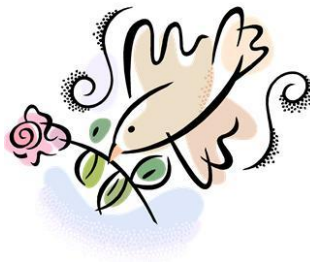
2 Brianna Jennings  
3 Brianna Ater  
10 Isabella Lowe  
    Ula Domi  
12 Roselyn Haire  
    Isabelle Sheahan  
13 Sarah Weber  
15 Dexter Robinson  
22 Chase Barno  
23 Ilan Betzer  
24 Peter Hopkins  
27 Naumir Osman  
    Alexander Clark  
29 Kyarah Moore  
31 Bailey Dunn

### November

4 Levi Berg  
    Paulina Adriano  
9 Taysen Wolfe-Saunsoci  
13 Cooper Heberlein  
14 Justin Pires  
17 Michael Passerino  
22 Abel Nichols  
28 Laken Holt

### December

2 Gregor Warmuth  
3 Alexis Hager  
4 Lillian Pruitt  
    Jaiden Robbins  
18 David Wooten  
20 Olivia Freeth  
29 Ethan Sargeant



## **STAY CONNECTED**

*Do you have questions and/or need to contact someone?*

### **Board of Directors**

#### **President**

Shannon Butalla  
shannon@thegfpd.org

#### **Vice President**

Anne Park Hopkins  
annepark@thegfpd.org

#### **Treasurer**

Melissa Bryce Gamble  
melissa@thegfpd.org

#### **Secretary**

Geoff Edgar  
Geoff@thegfpd.org

#### **Medical/Scientific Liaison**

Mousumi Bose  
mousumi@thegfpd.org

#### **Registry & Support Group Coordinator**

Pamela Marshall  
Registry@thegfpd.org

Woody Woodbury  
woody@thegfpd.org

### **US Regions & Regional Coordinators**

#### **New England & Mid-Atlantic (ME, NH, VT, MA, RI, CT, NY, PA, NJ)**

##### Coordinators

Jim & Jenn Charbonneau (VT) - vtshopper@yahoo.com, jenncharb@yahoo.com  
Jen Thompson (CT) - JenThompson82@yahoo.com  
Mousumi Bose (NJ) - mousumibose77@gmail.com  
Gina Bergh (NY) - ginababes@optionline.net, cheyennebabes@optionline.net

#### **South Atlantic (DE, MD, D.C., VA, WV, NC, SC, GA, FL)**

##### Coordinators

Jen Menard (NC) - jenmenard3@yahoo.com  
Katie Sacra (SC) - katsfsd@gmail.com

#### **North Central (WI, MI, IL, IN, OH, MO, ND, SD, NE, KS, MN, IA)**

##### Coordinators

Jeanine Sheppard (IL) - jsheppard@okawvillek12.org  
Patty DelSorbo (OH) - delsorbo@att.net  
Shannon Butalla (NE) - shannon@thegfpd.org  
Christina Leonhardt (SD) - cjleonhardt@spe.midco.net

#### **South Central (KY, TN, MS, AL, OK, TX, AR, LA)**

##### Coordinators

Bethany Barno (KY) - bethany.smelson@yahoo.com  
Liz Castaneda (TX) - ejchristian1@hotmail.com  
Jennifer Murphy (TX) - jenm1206@gmail.com

#### **Mountain (ID, MT, WY, NV, UT, CO, AZ, NM)**

##### Coordinators

Joleen Burdick (AZ) - jernjoghana@yahoo.com  
Faith Haire (AZ) - faithhaire@msn.com

#### **Pacific (AK, WA, OR, CA, HI)**

##### Coordinators

Heidi Harris (WA) - haharris@gmail.com  
Tracey Partridge (WA) - traceyp101@gmail.com

**We have a great online community. If you haven't plugged in yet, please consider doing so.**

[GFPD Family Support](#) (Parents & Guardians only)

[PBD Dads: A Band of Brothers](#) (Dads only)

[GFPD Shutterfly Site](#) (Parents & Guardians only)

[GFPD Extended Family Support](#)

[GFPD "Public" Facebook Page](#)

[GFPD website](#)

If you have recently moved and/or your information has changed, please email Pamela Marshall at [registry@thegfpd.org](mailto:registry@thegfpd.org) so we can update our records.

## **Medical & Scientific Advisory Board**

Dr. Gerald Raymond, M.D.

Dr. Nancy Braverman, M.D.  
McGill University  
Nancy.braverman@mcgill.ca

Dr. William Rizzo, M.D.  
University of Nebraska Medical Center

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Kennedy Krieger Institute

Dr. S. Ali Fatemi, M.D.  
Kennedy Krieger Institute

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University of Southern California

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Massachusetts General Hospital

Dr. Phyllis Faust, M.D., Ph.D.  
Columbia University Medical Center

Dr. Edward Kaye, M.D.  
Genzyme Corporation

**The GFPD is truly a global organization representing over 200 families from more than 20 countries around the world.**

- Argentina
- Australia
- Belgium
- Brazil
- Canada
- China
- Denmark
- Egypt
- France
- Germany
- Iran
- Ireland
- Mexico
- The Netherlands
- New Zealand
- Portugal
- Romania
- Saudi Arabia
- Scotland
- South Africa
- Sweden
- United Kingdom
- Uruguay
- United States of America

Australia – [Australian Leukodystrophy Support Group; ASIEM](#)

Belgium – [ELA Belgium](#)

Canada – [The Garrod Association](#)

Germany - [APS](#)

Italy – [ELA Italia; SISMMME](#)

The Netherlands – [Axel Foundation; VKS \(Adults, Children and Metabolic Disorders\)](#)

Spain – [ELA Spain; AECOM](#)

The United Kingdom – [Climb: National Information for Metabolic Diseases; ALD Life](#)

## SUPPORT GFPD

Raise money for the GFPD when you search the internet and when you shop online!



Designate contributions to GFPD through your employer's United Way Campaign or through the Combined Federal Campaign for Federal employees. Contact Melissa Gamble [melissa@thegfpd.org](mailto:melissa@thegfpd.org) for more information.



### Plan a Fundraising Event in Your Community

Please contact Melissa Gamble at [melissa@thegfpd.org](mailto:melissa@thegfpd.org) for more info.

**Make a donation in honor of or in memory of a loved one** GFPD is a 501 (c)(3) tax exempt public charity incorporated in the state of Oklahoma. By investing in GFPD, you are supporting children and families faced with a diagnosis of a Peroxisomal Biogenesis Disorder-Zellweger Spectrum Disorder (PBD-ZSD) and assisting family members and professionals through educational programs, research, and support services.

All contributions to GFPD are tax-deductible (USA) in the year paid. Donations are accepted by mail or online at [www.thegfpd.org](http://www.thegfpd.org).

Please mail checks to:  
Global Foundation for Peroxisomal Disorders  
5147 South Harvard Avenue, Suite 181  
Tulsa, OK 74135

### Become a Conference Sponsor

Without financial support, many families will not be able to attend 2013 GFPD conference. Please consider one of our conference sponsorship packages that will help offset the cost of food, lodging, and travel for the families and scientists attending the conference.

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