



# Winter 2013 E-Newsletter

## From the President

Much was done behind the scenes in 2012 for The Global Foundation for Peroxisomal Disorders. Our leadership team continued their quest to get the word out about the children impacted by Peroxisome Biogenesis Disorder-Zellweger Spectrum Disorder (PBD-ZSD) and the professionals who treat and study them. Some of our efforts included:

- Mailing 175 information packets to pediatric specialists and NICUs across the United States and Canada, which has increased family referrals to the GFPD tenfold.
- Maintaining a GFPD, which currently includes more than 275 children in over twenty countries throughout the world.
- Expanding our online presence through [www.thegfpd.org](http://www.thegfpd.org), Facebook & Twitter. Currently, the GFPD manages a support group online for parents of children with PBD-ZSD, a group for extended family/friends of children with PBD-ZSD, The PBD Band of Brothers (dad's only) and our community page.
- Hosting a Family Support Conference in Orlando, Florida with 150 participants.
- Conducting patient consults with world renowned professionals for twenty-eight children with PBD-ZSD at the 2012 Conference.
- Sending our message to nearly one thousand friends and donors of the GFPD via our year-end newsletter. Information regarding the 2012 conference, events, and current research was articulated in this mailing.

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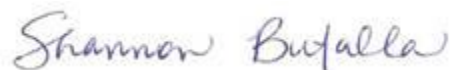
With the New Year, the momentum to grow the GFPD has increased. Numerous events in honor and memory of children living or deceased are happening nationwide, benefitting the efforts of the GFPD by supporting research, the 2013 GFPD Family & Scientific Conference, the equipment exchange program, and awareness efforts. Children diagnosed with PBD-ZSD make tremendous impact on the individuals in their lives, which is evident by the amount of support the GFPD has received in their honor.

Thank you to all who have helped the GFPD by donating their time, passion, and money to the organization. The GFPD is run completely by volunteers, who, most often, are parents of children with PBD-ZSD. We appreciate the willingness of parents to share their stories and experiences with others, as this information is often the most helpful for those facing a new diagnosis.

If you are the parent of a child with PBD-ZSD, we want to hear from you! Pamela Marshall, Registry & Support Group Coordinator, is gathering family stories to publish on the GFPD website. Please email Pamela at [registry@thegfpd.org](mailto:registry@thegfpd.org) for more details/instructions on how your child can be featured on the website.

We look forward to connecting with you this year! Please contact me if you are interested in getting involved, have questions about the GFPD, or want to share your story.

With hope,



Shannon Butalla

President, GFPD



The Butalla Family

## **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 Institute 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.**

## UPCOMING EVENTS

### First Annual Ilan-a-thon 5K Memorial Walk Run by Mousumi Bose



On April 21, 2013, we will be holding the First Annual Ilan-a-thon 5K Memorial Run/Walk benefiting the GFPD in Patterson Park in Baltimore City, MD.

This event will be held in memory of our son Ilan Betzer, who passed away October 2011 from the effects of PBD-ZSD at the age of 14 months. Registration for sponsorships and donations is now open at <http://ilanathon2013.eventbee.com>. Individual registration will open January 2013. All proceeds will benefit the GFPD. The race honoree for our First Annual Ilan-a-thon, will be T.J. Sacra, son of Katie and Ted Sacra. The Sacra Family are originally from Maryland and moved to South Carolina a few years ago.

Our location, Patterson Park, is one of the oldest parks in Baltimore, with a rich cultural and natural history, and just a few miles away from the Kennedy Krieger Institute. For registration and more information about Ilan-a-thon and other events supporting Ilan-a-thon, please visit <http://www.ilanathon.org>. To view the Ilan-a-thon video please click [here](#).



Andrew, Mousumi & Ilan

## SAVE THE DATE – 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.

Here are a few pictures from the 2012 GFPD Conference in Orlando, Florida.





## PAST EVENTS

### **Pound the Pavement for Peter – April 14, 2012 – Atlanta, GA**

The Global Foundation for Peroxisomal Disorders was very grateful to be the 2012 recipient of funds from *Pound the Pavement for Peter* and appreciative of the efforts by Peter’s Pals, the Atlanta/ Brookhaven community, and the family and friends of Peter Amann Hopkins.

In its third year, *Pound the Pavement for Peter* was a huge success with over 500 registered race participants, forty Corporate Sponsors, and forty- two Family Sponsors.

The 2012 *Pound the Pavement for Peter* Honoree was Jackson Jinright, a five-year-old Atlanta resident with a Peroxisome Biogenesis Disorder - Zellweger Spectrum Disorder.

Funds from the race will be used to enhance the lives of children with Peroxisome Biogenesis Disorder - Zellweger Spectrum Disorder (PBD-ZSD) and their families, as well as support the researchers passionate about finding treatments for this rare spectrum of disorders.



Angie Jinright, Jackson Jinright, Ashley Maple, Archer Maple, Anne Park Hopkins, Melissa Gamble, Sam Butalla & Shannon Butalla



## GFPD Dinner Dance – August 11, 2012 – Turlock, CA by Carolina Alfaro

On Saturday, August 11<sup>th</sup> we hosted our first GFPD Dinner Dance in memory of our two “angel” children Diego & Adrian to benefit the Global Foundation for Peroxisomal Disorders (GFPD). Both brothers were diagnosed with PBD-ZSD. My husband, Jose, and I made a promise to our boys before they died that we would keep their memory alive. To honor the memory of Diego and Adrian, we have made it our mission to host an annual fundraiser and give back to other children affected by PBD-ZSD and offer support to other PBD-ZSD families who are on this difficult journey.

The tri-tip dinner also included a silent auction, raffle and entertainment by “The Mike Torres Band” which had close to 250 guests dancing all evening. All food, beverage, silent auction and raffle donations were made by local businesses, friends and family. The event fundraised over \$15,000 for GFPD!

Everyone kept telling us that they felt the spirit of Diego and Adrian there that evening. Our special guests were Faith and Brandon Haire who also lost their daughter Roselyn to PBD-ZSD when she was six months old. Our guests were able to “physically” see that we were not the only family going through such grief. It was a very special evening.

The dinner dance was co-sponsored by: Swinerton Foundation, Sutter Health Tracy Hospital, State Farm Insurance, E&J Gallo Winery, F.G. Trucking Company, Eye Pro’s, DJ Exquisite Sounds & Lighting, Memorial Incorporated, Brown & Fesler, and Mistlen Honda.



Jose & Carolina Alfaro





## Art for Archer – October 20, 2012 – Meridian, MS by Ashley Maple

Our community of Meridian, Mississippi came together on October 20, 2012 for *Art for Archer*. The idea for this event came from my own home life with my children. I would save all of their artwork from the year, and then we would have a small art sale in our home for friends and family. We would then choose a charity to donate the money to. I wanted my children to know their art is valuable, and can help others in need.

*Art for Archer* showcased some of my photos, crocheted items, jewelry, art from my children, and many other donated arts and crafts. Finding creative outlets to channel my heartache has been essential to me. In a way, *Art for Archer* has allowed me to turn an ugly disorder and heartache into something beautiful and allowed me to do something for Archer and other children that share his disorder. You can read more about the event [here](#).



### Painting for PBDs – October 21, 2012 – Louisville, KY by Bethany Barno

To mark the one year anniversary of our son Chase’s death, we hosted “Painting for PBDs” at Uptown Art Uncorked in Louisville, KY. We wanted to do something special to celebrate his life and raise money for those who still struggle with these horrible disorders. Our event was a wonderful way to get people together to remember Chase, raise awareness for PBD-ZSD, and raise funds for GFPD.



### Spaghetti Dinner Benefit – January 27, 2012 – De Graff, Ohio

A spaghetti dinner benefit for GFPD was held on January 27, 2013 at the De Graff United Methodist Church in De Graff, Ohio. A silent auction accompanied the dinner. The event was held in memory of the Frost children. Leslie and Jerry Frost lost three daughters to PBD-ZSD: Angel, Rebekka, and Donna.

Despite the bad weather, many came out to show their support for the Frost family and to raise awareness of PBD-ZSD and funds for the GFPD.



J.J., Jerry, Leslie and Michael Frost

## **GFPD FAMILY NEWS**

### **Making a Difference for Children and Families at Leighton Hospital**

Laura Denny's daughter, Ellie, passed away from PBD-ZSD in 2011, but her memory lives on in the hearts of those that loved her and in the lives of those who are touched by the family and friends' fundraising efforts that are making improvements to the Children's Ward at Leighton Hospital. Read more on Laura's [blog](#).

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### **Paige Treen's Make-A-Wish Princess Room**

Paige Treen, the four year old daughter of Gail O'Brien and Peter Treen of Nova Scotia, Canada, recently received a princess room makeover from the Make-A-Wish Foundation. Read the full article [here](#).

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### **Habitat for Humanity Improves Accessibility for the Maxwell Home**

The Maxwell family of Tennessee recently had a number of home improvement projects completed by Unico County Habitat for Humanity which allowed for their home to be more functional for their daughter, Autumn. Read the full article [here](#).

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### **Sharing the PBD-ZSD Journey: Jim Charbonneau & Nancy Guthrie**

While many families utilize the Internet to keep family and friends updated about their PBD-ZSD journey and to spread awareness for this rare disease, two individuals have published books about their personal experiences.

James (Jim) Charbonneau is the author of *From Donor to Diagnosis: Raising a Special Needs Child*. Jim and his wife Jennifer are the proud parents of Ciera and Bailey. Their oldest daughter, Ciera, was conceived via donor insemination, and has PBD-ZSD. The book is available in paperback and electronic versions online at [Amazon](#). The Charbonneau family lives in Vermont.

Nancy Guthrie is an accomplished writer who has written two books, *Holding onto Hope*, and *The One Year Book of Hope*, about the loss of their two youngest children. Nancy describes her books: "*Holding onto Hope* is my journey through the book of Job in the Bible in the midst of losing my daughter, Hope, and my son Gabe, to Zellweger Syndrome. I wanted to figure out how this man lost so much, questioned God so boldly and yet emerged from it with a life described as 'good.' Because in the midst of that, I didn't think my life would ever be good again. *The One Year Book of Hope* is a one-year daily devotional for people who are hurting or grieving. Throughout its pages I work through the many passages of scripture that I found so difficult to understand and apply in light of my losses." Nancy and her husband David are also the hosts of Respite, a retreat for couples who have faced the death of a child. For more information on her books and/or Respite, please visit [www.nancyguthrie.com](http://www.nancyguthrie.com). The Guthrie family lives in Tennessee.

## Birthdays

### January

- 8 TJ Sacra
- 10 Ella Cook\*
- 11 Kyarah Moore\*
- 16 Danny Woodbury
- 18 Adrian Alfaro\*
- 21 Jonas Kruijff
- 23 Jydon Campbell
- 26 Zachary Metcalf
- 29 Roselyn Haire\*
- 31 Kenan Brown\*  
Jarrett Brown\*

### February

- 1 Gregor Warmuth\*
- 2 CJ Bergh
- 5 Emily Clifford  
Daniel Denis  
Nicole Pexito\*
- 7 Beau Olson\*
- 9 Fatimah Elreda\*  
Olivia Menard
- 12 Liam Dunn
- 13 Jamie Airington  
Theo Holmgard\*  
Zane Steward\*
- 14 Timothy Bishop\*
- 18 Casey O'Connor
- 19 Shane Robinson  
Jessica Watts\*
- 20 Skye Andrews
- 22 Autumn Maxwell  
Kayla Vondal\*
- 25 Thomas Goffaux\*  
Ashley Goffaux\*

### March

- 2 Maximus Manciu
- 3 Jackson Jinright\*  
Abel Nichols\*
- 5 Alyssa Vondal\*
- 6 Charlie Johnston\*
- 9 Ethan Sargeant\*
- 10 Hayden Kately
- 12 Kelsey Matt\*
- 14 Anastzia Hulkow
- 15 Alana Nguyen\*  
Zane Rock\*
- 17 Dylan Martin\*
- 19 Aden Crawford  
Ian Harris  
Christine Ragazzo\*
- 20 Abdullah Harak  
Naumir Osman\*  
Samuel Peacock\*
- 26 Andrew Patrick
- 27 Carter Leonhardt\*
- 28 Corey Sheppard\*
- 29 Sarah Weber\*
- 30 Alejandro Chavez  
Ana Paula Chavez
- 31 Anna Turci



\*deceased

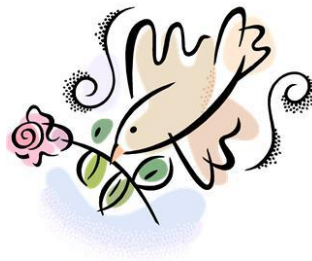
## Anniversaries

### January

- 4 Adrian Alfaro  
Parker Owens
- 5 Elisa Brechet
- 7 Emilia Clarke  
Christopher DelSorbo
- 8 Teagyn Smith-Hendricks
- 9 Caitlyn Hunter
- 12 Jessica Watts
- 14 Collin Brown  
Sofia Nadein
- 15 Gabriel Guthrie  
Gabriela Castaneda
- 16 Lillian Burns
- 17 Joshua Dilworth
- 18 Nicole Pexito  
Ross Phillipson
- 19 Kody Smith  
Khian Grant
- 21 Averi Owens
- 27 Lawson Hebert
- 31 Sienna Castro

### February

- 4 Jonah Johnson
- 9 Alec Reid
- 11 Vincent Carusone
- 17 Logan Diderrich
- 18 Alex Callaghan
- 22 Diego Alfaro
- 25 Thomas Goffaux  
Gwendolyn Hayes



### March

- 1 Harvey Jones-Langan
- 2 Isabella Frank
- 3 Ellie Prince
- 5 Jacob DeWitt
- 9 Alyssa Patrick
- 13 Lilly Dickison
- 15 Christine Ragazzo  
Natalie Morford
- 17 Alyssa Vondal
- 19 Franciscom Fernando
- 23 Joel Smith
- 25 Nicholas Braga
- 26 Zane Rock

### **The GFPD family mourns the recent loss of the following children due to the devastating effects of PBD-ZSD.**

- Brianna Ater**, April 17, 2012 – October 3, 2012 (North Carolina, USA)
- Sofia Martino**, June 20, 2012 – November 1, 2012 (Montivideo, Uruguay)
- Star Zheng**, 2009 - November 2012 (Guangxi, China)
- Millie McMenemy**, November 21, 2006 – December 1, 2012 (North Lanarkshire, Scotland)
- Jackson Jinright**, March 3, 2007 – December 7, 2012 (Georgia, USA)
- Tudor Proca**, June 6, 2011 – December 13, 2012 (Ilfov, Romania)
- Savanna Watts**, October 29, 2003 – December 29, 2012 (Kentucky, USA)
- Jessica Watts**, February - January 12, 2013 (Birmingham, United Kingdom)
- Sofia Nadeina**, November 29, 2012 – January 14, 2013 (Nebraska, USA)
- Lawson Hebert**, June 6, 2012 – January 27, 2013 (Louisiana, USA)

## **STAY CONNECTED**

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

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**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.

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

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

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

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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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