Rare Disease Day Campaign:

Our first official fundraiser for the Glut1 Deficiency Foundation was a great success! Together we raised just over $46,000! All of these funds will be put to work helping us in our mission of raising awareness, educating others, advocating for Glut1 patients and families, and supporting researchers as they work for a cure. We are investigating some exciting and promising research opportunities with plans to contribute significantly from these funds. We will be sure to share with all of you the projects our Board ultimately chooses to support and the amount of those donations.

We have many aspirations for the future of the Foundation, but more importantly, for the patients with Glut1 Deficiency. We are so grateful to all of you who participated and donated. Please visit our website to learn more about Glut1 Deficiency and how your contribution will help us with our ongoing and future projects.

The success of our fundraiser is further proof that we can do so much more together than any of us can do alone. Together, we can find the answers that bring quality of life to Glut1 Deficiency patients and families, and ultimately, find a cure.

Website Redo: Our new, improved, and enhanced website is nearly complete. We hope to debut the new site in the next couple of weeks and will be sure to let you know when all is ready. Our web address will remain the same.

www.g1dfoundation.org
We are pleased to present the 4th Annual Glut1 Deficiency Conference in beautiful downtown Indianapolis! Our location at the Embassy Suites (Group Code: GLT) is within walking distance to many downtown attractions, including the Indianapolis Zoo, Indianapolis Children’s Museum, Circle Centre Mall, and much more!

We are excited to report that Dr. Jörg Klepper from Germany will be a presenter this year to give us his report on the European experience with Glut1 Deficiency. We are also thrilled that Dr. Darryl De Vivo and Dr. Juan Pascual will be joining us again, along with members from their teams at Columbia University Medical Center and UT Southwestern Medical Center.

Our annual conference gives you the opportunity to meet new friends, share your experiences with old friends and learn what is new in the world of Glut1 Deficiency, as we all learn more and take steps that move us closer to a cure.

We look forward to seeing you there!

Please click here to register for the conference, order conference T-shirts, and make your hotel reservations.

The Hollemans have provided an outline of the conference and it is provided on the next page. An agenda is being finalized and will be shared as soon as it is all set.
Conference Agenda
please check our website links for updates as the agenda is finalized

Thursday, July 12, 2012

Early Afternoon:  Welcome and Introduction

Afternoon:  Presentations, followed by Meet and Greet with the Doctors

Informal discussion groups with all of the available presenters. A chance for you and your family to talk one on one with other families and Glut1 experts.

Evening:  Social Hour at the hotel

Friday, July 13, 2012

Morning:  Research Doctor Presentations

Lunch Break

Afternoon:  Break Out Sessions

5PM  Social Hour at the hotel

Evening:  Cultural Activity (Optional)

children’s activities and supervision provided throughout conference sessions
The Wienk family (Cody, Heather, and Haley) represented the Glut1 Deficiency Foundation at Sanford’s 2nd Annual Sanford Rare Disease Symposium on February 25th, 2012 at Sanford Research in conjunction with Global Rare Disease Day.

The goal of the Rare Disease Symposium was to provide information for healthcare professionals, researchers and the local community about current research efforts into rare diseases here at Sanford Research and nationwide. Guest speakers included Dr. John Shoffner of Medical Neurogenetics LLC (Mitochondrial Disease), Dr. Kate Rauen of University of San Francisco California (RASopathies), and Dr. David Robertson of Vanderbilt University (Multiple System Atrophy).

Other presentations included Dr. Michael Kruer, pediatric neurologist at Sanford Health and Associate Scientist in the Sanford Children's Health Research Center (Neurodegeneration with Brain Iron Accumulation); Dr. Chun-Hung Chan, Director of the Sanford Health Biobank and Liz Donohue, Director of the Coordination of Rare Diseases at Sanford (CoRDS) registry.

Nearly 175 people registered to attend the event in person and 25 registered to attend the event via webinar. Webinar attendees were from across the United States and from the United Kingdom, Czech Republic, India, Canada and Mexico.

Nine organizations attended the Symposium to host vendor booths to share information on how they support patients who have been diagnosed with a rare disease and their families.

Please click [here](#) for a summary report of each presentation.

**A huge thanks to the Wienks!**
Matt Golinski update:
We mentioned Matt in the last newsletter. His wife and three daughters perished in a house fire in Australia the day after Christmas, and Matt suffered life-threatening burns. Matt’s twin daughters had Glut1 Deficiency.

A local reporter has been sharing updates with us and she sent a link to a recent story with news about his healing progress, as well as information about how to donate to a recovery fund that friends and family have set up for him.

Click here for the link to the story.

Awesome Advocate

Olivia Guziewicz of New Hampshire recently had the opportunity to give a talk to her class at school about Glut1 Deficiency.

Here’s what her mom, Elizabeth, had to say:
She has been studying DNA, genes and mutations in 7th grade science class and Olivia wanted to speak about Glut 1. She touched on how rare it is, how it impacts her life and the special diet she is on. She said that because she has Glut 1 she is very special but is also just a normal kid! The class was SO receptive! They thought it was cool that she is one of only about 300 cases, that she gets to travel (sometimes by plane and by limo) to conferences and that she met Dr. De Vivo (he is famous) and has a pic with him! They also thought it was cool that she can eat hot dogs, pepperoni, sausage, whipped cream and butter! We are so very proud that she wanted to share with her class and are so very blessed that her class is so accepting and protective of her.

and this is what Olivia shared with her class:
GLUT1 is a genetic condition.
GLUT1 is a mutation of a gene that affects the brain.
GLUT1 is a rare disorder.
Less than 300 people in the world have GLUT1, I am one of those people.
We have to be on special diets so we can stay healthy and strong.
I have spent time with the Doctor who discovered GLUT1 in New York.
His name is Dr. De Vivo, he is famous! Because I have GLUT1, I am famous too!
Both girls and boys can have GLUT1.
I know 30 other kids with GLUT1.
My best GLUT1 friend is Macie.
We have spent time together in Chicago and in Kentucky.
Because I have GLUT1 I get to travel a lot.
I will travel to Indiana this summer to visit with my friends again.
The most important thing to remember about people with GLUT1 is that we are are just normal kids, with a very special gene!

Dylan’s story

Adam Watson and his mom Stefanie highlight the need for better understanding of Glut1 Deficiency in a story from Arbroath, Scotland.

Adam’s story

Matt Golinski update:
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watch a heartwarming and inspirational interview with Phil Parker, from England, on what it is like living with Glut1 Deficiency and the ketogenic diet:

Phil’s interview
Fundraising Families

Vivian’s talented and compassionate mother, Rachel Kuyper of Spring, Texas, is conducting a special fundraiser - CRAFTING A CURE FOR GLUT1. Head over to Belle Bébé Boutique on Facebook for great baby gifts! 100% of net proceeds go to support the GLUT1 Deficiency Foundation. Help "Craft a Cure for GLUT1"!
http://www.facebook.com/BelleBebeBoutique

Thank you Rachel and Vivi!

The Dean Family - Chris, Nichole, Jayden
Olive Hill, Kentucky

Their friends and family helped host a Zumbathon to raise money during the ♥Some1 with Glut1 campaign. Many thanks to the Dean family for a great event - raising awareness, educating others, and helping raise funds!

Milestones for Children held a successful fundraiser in conjunction with Rare Disease Day - Together To 100k. Milestones was founded by Glut1 mother Shelly Allen and raises money to fund Glut1 research at the Colleen Giblin Laboratory. She is a tireless and dedicated advocate for our children. Thank you Milestones for Children!

www.milestonesforchildren.org

please send future newsletter items or additions to mailing list to: info@g1dfoundation.org
Genetic Counseling

Kris Engelstad, from Columbia University Medical Center, recently received an MS in Genetic Counseling. She is available to speak to patients and/or family members regarding genetic issues in Glut1 DS. As always, she is also available to discuss general topics in Glut1 Deficiency. She can be reached at: 1-212-305-6834 phone or e-mail ke4@columbia.edu

Glut1 Patient Online Community

Thanks to NORD and Eurordis for sponsoring the Glut1 online community. This is a great resource for anyone looking for more information on Glut1 Deficiency and wanting to reach out to others sharing the journey.

The site is available in German, English, French, Spanish, and Italian, and you can request human translations for any posts. Please consider stopping by, registering, and even sharing your child’s story.

Glut1 DS, the Community

Rare Disease Communities

www.rarediseasecommunities.org

Resources

Glut1 Research Lab Links

UT Southwestern Rare Brain Disorders Clinic and Lab link

Columbia University Colleen Giblin Lab link

Recent Publications

(GLUT1 deficiency: A glut of epilepsy phenotypes

Growth hormone deficiency: a possible complication of glucose transporter 1 deficiency

GLUT1 mutations are a rare cause of familial idiopathic generalized epilepsy

Derivative chromosome 1 and GLUT1 deficiency syndrome in a sibling pair

Yahoo Support Group

If you are not currently a member, please consider joining the Yahoo Health Glut 1 DS Group. It is a wonderful way to communicate with and learn from other Glut1 families from around the world.

http://health.groups.yahoo.com/group/GLUT1DS

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please send future newsletter items or additions to mailing list to: info@g1dfoundation.org
Glut1 Deficiency Foundation

Mission:
The Glut1 Deficiency Foundation is a volunteer, non-profit family organization dedicated to:

* educating others about Glut1 Deficiency by creating a forum for sharing support, experiences, resources, and information between patients, families, and healthcare professionals.
* increasing awareness of and advocacy for Glut1 Deficiency.
* supporting and funding researchers as they work for a cure.

www.g1dfoundation.org

A Good Way to Help

Help support the Glut1 Deficiency Foundation by registering to use these free and easy programs:

GoodSearch: earn donations for web searches (powered by Yahoo)

GoodShop: earn donations through online shopping from your favorite retailers

GoodDining: earn donations by dining out at your favorite restaurants

Glut1 Deficiency Foundation Officers

President:  Glenna Steele - mom to Macie Owingsville, Kentucky gsteele@g1dfoundation.org

Vice-President:  Lloyd Holleman - dad to Chris Carmel, Indiana lholleman@g1foundation.org

Secretary:  Greg Stoddard - dad to Dalton Evansville, Indiana gstoddard@g1dfoundation.org

Treasurer:  Jen Lazar - mom to Sam Frisco, Texas jlazar@g1dfoundation.org

Member Representative:  Keri Meyers - mom to Katie Denham Springs, Louisiana kmeyers@g1dfoundation.org

THANK YOU
to all who are helping support the mission of the Glut1 Deficiency Foundation through your donations and fundraising. Your efforts are helping ensure a brighter future for all of our children.

mailing address and donations:
Glut1 Deficiency Foundation
PO Box 943
Westfield, Indiana 46074-0943
USA

online donations click here

The Glut1 Deficiency Foundation is a 501 (c)(3) non-profit organization

Don’t forget to check to see if your employer has a matching funds program for charitable donations - a great way to at least double your efforts!