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.

Foundation Updates

Planning Meeting: We were most fortunate to have the opportunity to meet with Dr. De Vivo, Dr. Pascual, Kris Engelstad, and Shelly Allen (Milestones for Children) during the New Orleans conference. We were able to get invaluable input in helping us plan for future projects as we move forward with our mission.

Brochures: Our brochure project is going strong. We have mailed packets of our informational brochures to neurology centers across the country. We also had the wonderful opportunity to distribute brochures at our exhibit booth at the Child Neurology Society Annual Meeting in Savannah October 25-27. We also have a booth reserved at the American Epilepsy Society meeting in Baltimore in December, and are hopeful to get a spot at the American Academy of Neurology conference in New Orleans in April.

Glut1 Deficiency Video: we created an informational video about Glut 1 Deficiency for use as a tool to help raise awareness and educate others. We featured the video at our booth at the CNS conference, and it is posted to YouTube with links on our website. If you haven’t watched, please check it out at: http://www.youtube.com/watch?v=nzPusEZeq-A

We are hopeful that some major advancements will bring about the need for a revised version of the video in the not-too-distant future. If you didn’t submit a photo or video of your child this time, please consider doing so for future versions.

Future Projects: A major project we want to be involved with in the near future is the creation of a patient data registry for Glut1 Deficiency. We are also hopeful to help facilitate the creation of a Uniform Standard of Care document.

Rare Disease Day: Mark your calendars for February 29, 2012 - Rare Disease Day across the world. We will be involved in a joint project to help raise awareness and funds for Glut1 Deficiency on Rare Disease Day - watch for more details to come.

Conference: Our New Orleans conference was a huge success. We had 34 different Glut1 Deficiency families in attendance - 17 new and 17 returning. You may find our conference summary and lots of photos at our website. Plans are well under way for the 2012 conference in Indianapolis July 12-13. We hope all of you can join us!

Please watch our website for conference updates: www.g1dfoundation.org

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@g1foundation.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

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!
The third annual Glut1 Deficiency Conference was held July 14-15th in New Orleans, Louisiana. 34 families from around the world attended, where we had the opportunity to meet others, share experiences, and learn the latest from the experts. It was an unforgettable experience on many levels, and we were given many reasons to be hopeful and encouraged about the future for our children. We are grateful and most fortunate to have such dedicated and compassionate specialists working to help them. Many thanks to the Meyers family for hosting and to all who were able to join us. You may find a link to the summary of the conference presentations on the home page of our website: www.g1dfoundation.org.

THE KIDS HAD A BLAST!

Yes, that is a real alligator puckering up with Pat Hemlock - Drew’s mom.

Yes, we brought a horse INSIDE the hotel....

and yes, that is a horse AND a clown hanging out in the hotel bar.

many more photos on our website
The Glut1 Deficiency Foundation was honored to have the opportunity to participate in the 40th annual meeting of the Child Neurology Society in Savannah, Georgia. The Stoddard family (Greg is a Foundation board member/officer) represented us at the exhibit hall, where Greg, Debbie, and Dalton had the chance to talk with many pediatric neurologists about Glut1 Deficiency. This was the first of what we hope to be many more opportunities to educate and raise awareness among medical professionals through conference exhibits.

April Breen will represent the Foundation at the 65th American Epilepsy Society meeting in Baltimore December 2-6.

We also hope to secure a nonprofit booth space at the 64th annual American Academy of Neurology meeting in New Orleans in April 2012.

Other upcoming related conferences:

International Symposium for Dietary Therapies for Neurological Disorders:


Many of our Glut1 Deficiency families have recently participated in a study at UT Southwestern where a specialized MRI machine is being used to measure brain metabolism (see Chris and Lloyd Holleman at left).

The study has just wrapped up, and Dr. Pascual reports that the research team obtained “unprecedented data”.

Dr. Pascual is also anticipating the start of the C7 oil (triheptanoin) study in the very near future. Minor changes are being made and IRB approval is expected at any time. Glut1 Deficiency families, led by the Meyers family and with the help of The Child Brain Foundation, raised the funds for the C7 study.

You may find more information about each of these studies in our conference summary.
Genetic Counseling

Kris Engelstad, from Columbia University, 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

New Glut1 Deficiency Resource:
Wonderful, brand new information on Glut1 Deficiency (G1D) made possible by the collaboration between The Child Brain Foundation, Dr. Juan M. Pascual, and The National Institute of Health’s Office of Rare Diseases Research (Collaboration, Education and Test Translation Program).

You may also download a PDF version of the information from the website.
www.childbrainfoundation.org

Glut1 Deficiency Grand Rounds at Cincinnati Children’s Hospital Medical Center

Dr. Juan Pascual “Brain Energy - Excitability Diseases”
August 3, 2011

Macie and Glenna Steele (Foundation board member/officer) were in attendance for some of the sessions and were able to distribute our brochures.

Grand Rounds are an excellent way to raise awareness and educate the medical community about Glut1 Deficiency. Talk to your healthcare center to see if they are interested - a personal connection to a patient(s) with a particular condition often helps generate interest.

New Glut1 Patient Online Community

Just launched - the Glut1 Online Community! Thanks to NORD and Eurordis, this will be a wonderful 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. Please consider stopping by, registering, and even sharing your child's story.

Glut1 DS, the Community Rare Disease Communities
www.rarediseasecommunities.org

in the news .... recent Glut1 Deficiency publications (abstracts only)

Paroxysmal choreoathetosis/spasticity (DYT9) is caused by a GLUT1 defect
Glucose transporter type 1 deficiency syndrome with carbohydrate-responsive symptoms but without epilepsy
Glut1 Deficiency: When to suspect and how to diagnose
SLC2A1 gene analysis of Japanese patients with glucose transporter 1 deficiency syndrome
Dear Sir/Madam,

I am writing to invite you and/or your child to participate in a new initiative here at the Sanford Children’s Health Research Center (SCHRC) which is part of Sanford Health in Sioux Falls, SD. The newly established SCHRC shares a unique partnership with the Sanford-Burnham Institute for Medical Research (La Jolla, CA), as well as national and international research relationships through Sanford Children’s Hospital. The mission of the SCHRC is to develop translational research for rare diseases to drive the development of new therapies from discoveries made in research labs by integrating basic science with clinical practice, a so-called “bench-to-bedside” approach.

Many new developments in the diagnosis and treatment of human disease are the result of research that involves the collection and analysis of medical records of patients with a certain disease or condition. Unfortunately, one major limitation for research on rare diseases is the relatively small number of patients from which study participants can be recruited. As you may be aware, a rare disease is considered to affect less than 200,000 individuals in the US, although many rare diseases number in the low thousands, hundreds, or even only a handful. The small number of patients can cause problems in identifying common themes in disease, and presents challenges in the identification of individuals eligible and interested in participation in clinical trials.

In an effort to achieving the aforementioned new development and further understanding of rare disease we have established the Coordination of Rare Diseases at Sanford (CoRDS) registry. The purpose of the CoRDS registry is to facilitate research on rare diseases by providing a resource through which researchers can screen for prospective study participants. Any researcher in the United States who is actively engaged in research on a rare disease would have access to this registry following consultation with a CoRDS advisory panel. This will help accelerate rare disease research by providing a readily available registry of patients who may be eligible for participation in future clinical trials.

For more information and to learn how you can enroll, please visit our sites: [www.sanfordresearch.org/CoRDS](http://www.sanfordresearch.org/CoRDS) and ‘Sanford CoRDS’ on Facebook and Twitter.

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**Rare Disease Video Project**

Dr. Wendy Chung, a genetic specialists from Columbia University, would like one or two families who are willing to tell their story about their Glut1 child. What they recognized as different in their child, the diagnostic journey, the treatment and how it has worked, and what effect getting a diagnosis has had. Interviews have typically been done at Columbia in New York, so any area families should consider participating. If you are interested but can’t make it to NYC, then the taping could be done elsewhere. Please contact Dr. Chung for more information.

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**Conference T-Shirts**

Please submit your child’s artwork for our 2012 Conference T-shirt project. A Glut1 child’s art will be chosen and featured on our conference T-shirts for next year. We tried out the idea in New Orleans and it seemed to be a big hit. We will list sponsors on the back of the shirts next year along with our logo. Even though only one piece of art will be chosen for the shirt front, we would like to use all submissions to create a keepsake project of some kind - perhaps a calendar or greeting cards. We hope many of you will participate.

Please scan artwork and email to Lloyd Holleman at: lholleman@g1dfoundation.org

or you may mail the original to him at our foundation address:

Glut1 Deficiency Foundation  
PO Box 943  
Westfield, Indiana 46074-0943  
USA

Deadline is March 31st

You can see this year’s design below to give you an idea:

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**Help Wanted**

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PO Box 943  
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Deadline is March 31st

You can see this year’s design below to give you an idea:
Hello Families!

For the Glut1 Deficiency conference held in New Orleans this past summer, I had some silicone wristbands made up to give away as gifts to all our Glut1 families. My slogan on the wristband says "I LOVE SOME-1 with GLUT-1". I made a lot of them and I want to share them with all our Glut1 families!

If you are interested in having some of these wristbands for your family (and I really hope you are) I would be happy to send some to you at no charge. I'll cover both the wristband cost and the postage (even overseas.) I love the idea of every child and every family member touched by Glut1 having a special wristband that is all ours and that in a small way helps to unify us all. I will send the wristbands out to anyone in the Glut1 community who asks for them. Just email me at gstoddard8@insightbb.com and I'll get them out to you as quickly as I can.

Dalton is pictured holding a handful of them and has so enjoyed sharing these wristbands with family and close friends. I wear mine all the time, as does our entire family. I hope you'll enjoy wearing them too! Warm wishes to all, especially as it begins to chill-down here!!!

Debbie Stoddard, mom to beautiful 14 year old Dalton

Fun 4 Glut1

A huge thank you to the Holleman Family (Lloyd is a Foundation board member/officer) for hosting a wonderful fundraiser in September to benefit the Glut1 Deficiency Foundation and our 2012 conference effort.

Please visit the event website to see more about this great event - attended by 3 Indiana Glut1 families!

www.fun4glut1.org

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/

Newsletter Submissions

We aim to make this a quarterly newsletter. If you have items or information to submit, or ideas or suggestions for inclusion in future issues, please email to:

gsteele@gl1dfoundation.org

National Walk for Epilepsy

Please be thinking about the possibility of a Glut1 Deficiency team at the 6th annual National Walk for Epilepsy in Washington, DC on March 31, 2012. If there is enough interest from enough families who could come, it would be a great opportunity for education/awareness as we walk along the National Mall...not to mention great fun and fellowship! Let us know what you think.

www.walkforepilepsy.org