Glut1 Deficiency Foundation

Newsletter

volume 4
Winter 2012

Foundation Updates

Official Non-Profit Status: We were thrilled to receive our official non-profit 501 (c)(3) designation from the IRS in November (retroactive to July 2011). This was a big step for our Foundation and will open many doors in helping accomplish our mission. Lots of time and energy went into the application process, and we were fortunate to have the volunteer services of two attorneys who were meticulous and thorough and helped our application get approved on the first attempt. Many thanks to all who have helped see this dream realized.

Rare Disease Day: Mark your calendars for February 29, 2012 - Rare Disease Day across the world. We will be having a special fundraising and awareness campaign for the entire month of February, culminating on Rare Disease Day. Please see the information on page 2 about how you can be a part of this campaign - it’s easy and we need you! Those families on the mailing list will receive information about participating via email.

2012 Conference: This year’s family medical conference will be hosted by the Holleman family in Indianapolis, Indiana on July 12-13. The Hollemans are working on venue and speaker/topic plans, and we have participation from Dr. De Vivo, Dr. Pascual, Dr. Klepper and their team members along with other presenters with topics of importance to Glut1 families. We will share updates on our website and our future newsletters. We hope you’ll be able to join us! This year, we will be offering online registration, which we hope will be a help and a convenience to you. Registration will open soon, so watch for more details to be shared.

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 experience of distributing brochures at our exhibit booth at the Child Neurology Society Annual Meeting in Savannah in October and at the American Epilepsy Society meeting in Baltimore in December. Our next opportunity will be the American Academy of Neurology meeting in New Orleans in April - with a projected attendance of 14,000 health care professionals from around the world.

Website Redo: We are in the process of revamping our website to make it a better and easier tool for sharing information and resources. We expect to debut the new site in March and will be sure to let you know when all is ready. Our web address will remain the same and our current site will remain available throughout the process.

www.g1dfoundation.org
Calling All Glut1 Patients!

The Glut1 Deficiency Foundation is conducting our first annual fundraiser in conjunction with Rare Disease Day and we need your participation! While the Glut1 Deficiency Foundation is located in the United States, we support projects that will help all patients, all around the world. We hope that you will join us in our efforts to continue working on our mission of education, awareness, advocacy, and supporting researchers as they work for a cure.

Dr. Pascual and Dr. De Vivo have several promising research projects on the horizon and we can all benefit from their success. Visit our website (www.G1DFoundation.org) to see all of the projects - large and small - that we have already accomplished, those that are ongoing, and those that we plan for 2012. We need money to keep these programs going. Please consider having your Glut1 child participate as a fundraiser in this important campaign!

**WHO:** All Glut1 patients, families, friends or supporters

**WHAT:** Patients will raise money to help fund the upcoming projects of the Foundation through the solicitation of donors. Each patient will have his/her own website to share with family, friends and supporters. The website will track donations and the progress towards a fundraising goal. Our goal is for each patient to raise $1,000, although there is no limit to the amount you may raise and no donation amount is too small.

**WHEN:** The campaign will run for the month of February, ending on National Rare Disease Day, February 29, 2012. In order to launch the campaign by the beginning of February, each participating patient will need to complete the below registration by **January 22**. This will allow the Foundation time to build your personal website. During this time, you should begin compiling your list of email addresses for solicitation.

**CONTACT:** Jennifer Lazar, Glut1 Deficiency Foundation, email: JLazar@G1DFoundation.org

**REGISTRATION:**

- Fundraiser (Patient) Name:
- Fundraiser (Patient) Birthdate:
- Contact Email address:
- Picture in .jpeg format:
- Personalized paragraph (100 words or less):

Upon receiving the above information, the Foundation will create your personal website and send you the URL address that you can email to your list of potential supporters. **Our success depends on you!**

See examples: 1  2  3
In December, April Breen (mom to Tessa), represented the Foundation as an exhibitor at the American Epilepsy Society’s (AES) Conference in Baltimore, MD and provided this summary: The conference was well attended by almost 4,000 neurologists, epileptologists and researchers from all over the world. We had excellent booth placement and exposure on the main walkway (attendees had to pass our booth to get to the food; so you can be sure they passed by several times!). Many attendees stopped to talk more about Glut1 and our Foundation, and to pick up our brochures and research articles. Several neurologists said they were already aware of our Foundation from their patients’ families, which was great to hear! With everyone’s help we’re spreading the word and raising awareness!

As mentioned in the last newsletter, the Stoddard family represented the Glut1 Deficiency Foundation at the Child Neurology Society annual meeting in Savannah, GA in October. Greg has provided a summary of their experiences: We were made to feel very welcome by the organizers and all of the participants. We received a number of positive comments about our booth, logo, and the fact that we were present. There were approximately 1000 attendees from all over the world, 900 of which were MDs (mostly Pediatric Neurologists). Our booth was visited by about 100 attendees and we passed out 60 review articles and over 200 brochures. Over 95% of our visitors were well aware of Glut1 Deficiency which was very encouraging. Many were actively testing for it in their practice and about 30% currently worked with Glut1 Deficiency patients. One group alone had identified eight Glut1 Deficiency patients. Clearly increased awareness, expanding phenotypes linked to Glut1 Deficiency and more active screening by doctors are leading to a significant increase in identified cases. The level of testing and number of increasing diagnoses was a positive surprise to us. We visited most of the Genetic testing booths and were impressed to find that nearly all included a Glut1 Deficiency screen as part of a standard assay package for many symptoms. Dr. De Vivo stopped by our booth a couple of times and discussions with him were a highlight of our trip. We are very fortunate to have such an outstanding researcher and advocate for this condition. In general we felt that there was tremendous momentum in this community related to Glut1 Deficiency and that we were there at the right time. We strongly recommend that the Foundation continue to support attendance of these conferences.

We will also have booth space at the 64th annual American Academy of Neurology meeting in New Orleans in April 2012.
New Article from Dr. De Vivo team
at Columbia University Medical Center, highlighting the use of the red blood cell glucose uptake assay as a diagnostic test for Glut1 Deficiency. Dr. De Vivo kindly provided us an introduction to the article:

"Transport of glucose across the red blood cell membrane provides us with a glimpse of glucose transport across the blood-brain barrier. To this extent, the red blood cell glucose uptake assay is a very reliable functional measure of glucose transport into the brain for nearly 100% of patients. For the moment at least, this assay is recognized as the diagnostic gold standard for Glut1 Deficiency Syndrome. When abnormal, we know that the patient has Glut1 Deficiency, even if we cannot find the disease-causing mutation."

Glut1 Deficiency Syndrome and Erythrocyte Glucose Uptake Assay

New Article from Dr. Klepper team
Glucose Transporter Type1 Deficiency Syndrome With Carbohydrate-Responsive Symptoms But No Epilepsy

Article from London
The Ketogenic and Related Diets in Adolescents and Adults--A Review.

Online Information from Dr. Pascual
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

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/

Glut1 Patient Online Community
Many thanks to NoRD and Eurordis, who have recently launched a 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

Glut1 Deficiency Video: We have over 1,000 views now of our Glut1 Deficiency video. If you haven't watched it yet, please follow the link:

Some of the Facts and Faces of Glut1 Deficiency

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

please send future newsletter items or additions to mailing list to: info@g1dfoundation.org
Adults and Diets for Epilepsy
Webinar from Johns Hopkins

Please join us for a webinar focused on adult dietary therapy for epilepsy, presented by the Carson Harris Foundation and hosted by Mackenzie Cervenka, MD and Bobbie Henry, RD, of the Adult Epilepsy Diet Center at Johns Hopkins Hospital.

Please join us for our next webinar: “Adults and Diets for Epilepsy”

Presenters:
Mackenzie Cervenka, MD & Bobbie Henry, RD
Adult Epilepsy Diet Center
The Johns Hopkins Hospital

Information:
Date: Tuesday, January 17, 2012
Time: 06:00 PM EST
Duration: 1 Hour(s)

In order to participate in the webinar you must pre-register at the following link:

https://carsonharrifoundation.linc.com/register/bfvrfks

Once you register, a “join” link will be sent to you. Please use this custom join link to attend the webinar.

Clinical Trial Update

Dr. Pascual reports that the C7 (triheptanoin) study has gained final approval this week pending some minor tweaking in the consent forms. He expects it to be ready to start soon.
The patient registry project will follow soon after, and the Glut1 Deficiency Foundation has committed to fund the registry project as well.

A website (TBA) is being created as a means to share information and direct questions about these and other projects at UT Southwestern. We will announce that website address as soon as it is ready.

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

go to www.goodsearch.com or contact the Stoddard family at gstoddard8@insightbb.com for instructions on setting up this simple and cost free way to raise funds for Glut1.

Glut1 Patient Story

A great story featuring Reed Williams and information about Glut1 Deficiency and the ketogenic diet.


please send future newsletter items or additions to mailing list to: info@glut1foundation.org
Condolences

Our heartfelt condolences go out to Matt Golinski, whose wife and three daughters died in a house fire a few days ago in Queensland, Australia. Matt also suffered severe burns and remains in critical condition. His 12-year-old twin daughters had Glut1 Deficiency. Matt’s family was instrumental in connecting Glut1 families in Australia and New Zealand.

A recovery fund for Matt has been set up by his friends and coworkers. We contacted the bank that is holding the account and asked about the process for making donations from overseas. Here is the reply:

Instead of sending a cheque, we recommend that you complete an International Money Transfer from an overseas bank account. You may need to provide the following -

1. The “SWIFT” code of ANZ (same as ‘SORTING’ or ‘ROUTING’ codes). = ANZBAU3M.
2. BSB Number - 014672
3. Account Number - 263331805
4. Account Name - Matthew Golinski recovery fund
5. The name of the bank - ANZ
6. The physical address of the bank - ANZ Noosa Heads
   23 Sunshine Beach Rd
   Noosa Heads
   QLD
   4567
   Australia

We also send our thoughts and best wishes to Dr. Juan Pascual, whose father passed away at home in Spain in November.

Help Wanted

Please submit your child’s artwork for our 2012 conference T-shirt project. We would like to feature the work of a Glut1 child or a sibling on the front, with sponsoring organizations, businesses, and families listed on the back.

Even though only one piece of art will be chosen for the shirt, 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

Upcoming Conference

International Symposium for Dietary Therapies for Epilepsy and Other Neurological Disorders

The Charlie Foundation/Matthew’s Friends

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!

Please e-mail Glenna by January 31st if you are interested:

gsteele@g1dfoundation.org

www.walkforepilepsy.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

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

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!