In our small Pennsylvania town there are two children with GSD 1a: our daughter Tessa and Jacob, son of Todd and Yolanda Hershey. As parents, we want the best for our kids and a cure for GSD is at the top of our list.

Tessa Peck & Jacob Hershey

Our families decided to have a fundraiser to help. On May 20, 2012, we hosted a Basket Bingo and Auction and donated all money raised to The Children's Fund. We had many beautiful themed Longaberger baskets filled with some really great prizes, donated items for auction, a bake sale and food stand. Our community came together and raised $7000.00. We would like to thank those who volunteered their time to help, those who came and played and spent the day with us and last, but not least, for the many donations we received. What a great day we had and with not one empty seat! -Mark & Melinda Peck

Cocktails for a Cure

The Bagin Family

Gary and Jennifer Bagin of Mullica Hill, N.J., raised over $31,000 for The Children's Fund for GSD Research at their March event in support of their son Justin, age 13 (1a). More than 250 friends and family attended "Cocktails for a Cure" at a local restaurant and generously bid for Chinese auction prizes and in a silent auction that included a 100-person pig roast, a week's vacation in Vermont, sports tickets and autographed jerseys, golf foursomes, overnight stays and more -- all donated by friends and family.

With mild coercion, Justin stepped up to the mic to thank everyone for their generosity. "We were completely overwhelmed by the love and support for Justin and GSD," said the Bagins.

Stephanie Samuelson, Rachel Messina, Cathy Trimble, Carole Bagin, Shannon Bray and Ashley Creta

Wine & Chocolate

Chocolations, Mamaroneck, NY

Jeanne Muchnick and Mark Rosen with Wendy Feldman

On March 14, 2012, an evening of wine, chocolate and fund-raising brought in more than $4,000 for The Children's Fund for GSD Research. Jeanne Muchnick and her husband Mark Rosen have put together this annual event in various forms for the past 4 years. This time, the event had something special -- appearances by Karen and Michael Gordon of Chappaqua, NY and Wendy Feldman of Cheshire, CT. She says it was the fact that two GSD moms came to support her -- and brought their friends that made the difference. Jeanne also wants to thank her friends Chris Ann Sepkowski, Melanie Rose, Katie Schlientz and Susie Sigel for their unwavering support as well as Chocolations owner Maria Valente who graciously donated her space. And of course, husband, Mark Rosen, for being her "rock."

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"...because every child deserves to be healthy."

The GSD Annual Super Bowl Raffle

Annual Super Bowl Raffle
Trip for Two to Super Bowl XLVII
New Orleans, Louisiana

Grand Prize Includes:

- Two tickets to Super Bowl XLVII in New Orleans, LA on 2/3/13 at Mercedes-Benz Superdome
- 5 days/4 nights accommodations at the Hilton Garden Inn (double occupancy) in New Orleans, LA
- Two playing spots in 8th Annual Super Bowl Celebrity Bowling Classic on 2/31/13 redeemable at All Star Lanes, Kenner, LA
- Two playing spots in 27thAnnual NFL Super Bowl Celebrity Golf Classic on 2/1/13 redeemable at Lakewood Golf Club, New Orleans, LA.

A total of 1000 tickets will be sold and tickets are $100 each.
The drawing will be held **Sunday, December 16th, 2012 at 5:00 PM**
For more information, please contact Teri Reed:
203-972-9199 or email treed04@gmail.com

Annual Super Bowl Raffle
Supporting The Children's Fund for GSD Research
Super Bowl XLVII
"...because Every Child Deserves to be Healthy."

"Going to the Super Bowl was most exciting thing I have ever done! I have always wanted to attend a Super Bowl, and having the opportunity to do so was amazing. The hotel was super fun too because a ton of previous Super Bowl players were staying there and it was great to be able to talk with them and see all the past super bowl rings. I think my favorite part of the whole trip (besides the game) was walking down the street in Indiana and seeing Joe Montana my all time favorite player!"

"It was not only my first Superbowl, but my first professional football game ever. I'm a big 49ers fan, but have always watched them on TV. It was just great being there . . . now I want to go to more games in person!"

Congratulations To Paul Rosenstiel, Our Super Bowl XLVI Raffle Winner!

Congratulations to Paul Rosenstiel of San Francisco, CA who won a trip to the Super Bowl XLVI. The winning ticket was sold by Mike Julius. Many thanks to those who purchased tickets and/or made donations. We sold 830 tickets and had many donations in addition to raffle tickets, so we raised in excess of $100,000. We are blessed to know so many caring and generous people. -Teri, Fran, Jack and Anna Reed.

Hopes and Dreams

The Newsletter for Friends of
The Children's Fund for Glycogen Storage Disease Research, Inc.

WINTER 2012/2013 GSD NEWSLETTER VOLUME 10

CONTACT US
917 Bethany Mountain Road
Cheshire, CT 06410
(203) 272-CURE (2873)

WEBSITE
www.cureGSD.org

E-MAIL
info@cureGSD.org

Mission Statement:
We are committed to funding research so that children born with GSD1 will benefit from early detection, treatment and an eventual cure.

2012 WAS MOMENTOUS For GSD Patients And Their Families... And YOU Helped Us Get There!

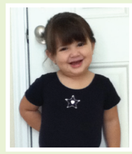
Finally a Full Nights' Sleep!

As a direct result of our first research symposium in 2004, research funded by The Children's Fund for GSD Research discovered a longer acting, more complex starch that extends the intervals for GSD patients between starch doses. We then funded studies to test this new "super starch" to determine the length of those intervals and proper dosing. After years of trials and perseverance, **we are thrilled to let you know that the FDA has approved a product called Glycosade, for the therapeutic management of GSD patients.** Patients can now safely last 8-10 hours overnight. This is one of the greatest advancements for Glycogen Storage Disease since the discovery of how cornstarch can be used to maintain glucose levels. Parents are now able to sleep through the night...a luxury that only a short time ago was only a dream.

THANK YOU!

A Cure for GSD-1

This box is not yet checked because we have not reached our ultimate goal of finding a cure for Glycogen Storage Disease, Type 1. We have your research dollars hard at work. We are gearing up for a 2014 human clincal trial using gene therapy. Inside this newsletter you can read about the amazing progress being done with the support of The Children's Fund. We will keep you posted as we get closer and closer and we keep our fingers crossed for a cure!



Montse Guerra

Jamie Gussin

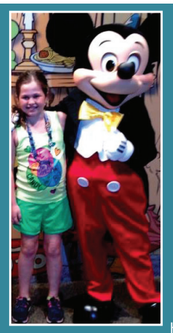
Jonah Feldman

Adam Julius

"...because every child deserves to be healthy."

SAVE THE DATE
7TH ANNUAL
GOLF CLASSIC & SPEAKEASY CLUB

FRIDAY APRIL 19, 2013
HERON BAY GOLF CLUB
CORAL SPRINGS, FLORIDA
BROUGHT TO YOU BY LISA & SANDY HODES



Thank You to Huntsville Elementary School for your continued support in honor of Rylee Graham!

Ryan Kalayci, Paige Hoss & Katie Hodes, selling GSD bracelets for The Children's Fund.

Abby Siegel, a good family friend of the Hodes girls made a lemonade stand to raise money for GSD research.

Thank You Norma and Ron Kahl of West Haven (dear friends of Anna Reed's grandmother Barbara Reed) who asked for donations in honor of their 50th wedding anniversary...over $750 was raised.

Anna Reed

Thank You to the NBPS Middle School Student Government for raising almost $900 for The Children's Fund last school year.

RESEARCH UPDATE 2012

National Institutes of Health
Janice Chou, Ph.D.

Patients with glycogen storage disease type Ia (GSD-Ia) are deficient in the enzyme glucose-6-phosphatase-α (G6Pase-α or G6PC). All patients manifest impaired glucose homeostasis while 70-80% of patients over age 25 years also develop long-term complications of hepatocellular adenoma (HCA). Our current therapeutic approach is to use gene therapy to restore G6Pase-α enzyme activity in the patient. To develop a safe and effective method for doing this we have been working with a mouse model of GSD-Ia. Our therapy works by putting the G6PC gene in a novel virus-based vector, named ssAAV-G6PC-GPE. When introduced into the GSD-Ia mice this virus delivers the gene into the liver and restores their G6Pase-α activity. The key characteristic of our approach is that we use both the natural human *G6PC* gene and the natural human *G6PC* gene control elements in our vector to ensure it expresses the G6Pase-α enzyme properly in patients when it reaches human clinical trials.

While restoring G6Pase-α enzyme activity is the key requirement in human clinical trials because it should rectify the life-threatening hypoglycemia characteristic of GSD-Ia. The best therapy would also prevent the longer term complications of HCA. To see if our therapy could address this, we treated GSD-Ia mice with varying doses of ssAAV-G6PC-GPE, designed to restore anywhere from 3% to 100% of normal liver G6Pase-α activity, and observed the impact of this therapy over 70-90 weeks. We found that all treated mice who restored more than 3% of normal liver G6Pase-α activity exhibited normal blood glucose levels, both before and after 24 hours of fasting. Importantly, none of the treated GSD-Ia mice developed HCA. Our results demonstrate that ssAAV-G6PC-GPE-mediated gene transfer in GSD-Ia mice not only corrects metabolic abnormalities for most of the natural life of a laboratory mouse, but that over their life there is no evidence for HCA formation.

Our vector, developed at NIH, is not the only vector that has shown good efficacy in resolving hypoglycemia in GSD-Ia mice. An alternative vector has been developed at Duke University. The major differences between the Duke vector (dsAAV-G6Pase) and the NIH vector (ssAAV-G6PC-GPE) are:

- The Duke vector is double stranded DNA (ds) while the NIH vector is single stranded DNA (ss)
- The Duke vector uses a smaller promoter called the *G6PC* minimal promoter (miGPE) at nucleotides-382 to -1; while the NIH vector uses a much larger promoter with more control elements in it called the human *G6PC* promoter/enhancer (GPE) at nucleotides -2864 to -1

In the current study Dr. Dwight D. Koeberl from Duke University and Janice Chou from NIH set out to conduct a side by side comparison of dsAAV-G6Pase and ssAAV-G6PC-GPE vectors, to determine which was the most efficacious vector for a clinical trial for the treatment of GSD-Ia. To ensure fair comparison, each vector was manufactured independently at the Powell Gene Therapy Center, University of Florida, and tittered both by the University of Florida and Duke University. The study consisted of performing gene therapy on 2-week-old GSD-Ia mice and monitoring phenotypic correction out to age 10-12 weeks. The results showed that NIH ssAAV-G6PC-GPE vector directed significantly higher levels of hepatic G6Pase-α expression and achieved greater reduction in hepatic glycogen storage than the Duke vector. The NIH ssAAV-G6PC-GPE vector-treated GSD-Ia mice could also tolerate fasting better than the Duke dsAAV-G6Pase vector-treated GSD-Ia mice. In conclusion, the study demonstrated that the NIH vector, ssAAV-G6PC-GPE is better than the Duke University vector dsAAV-G6Pase for the correction of murine GSD-Ia and therefore the NIH vector is the leading candidate for human clinical trials.

Duke University Medical Center
Dwight Koeberl, M.D., Ph.D.

Advances toward gene therapy with adeno-associated virus (AAV) vectors for GSD-Ia.

We completed two grants from the Children's Fund this year, and we wish to express our gratitude for this support of our research. Titles of these grants were as follows:

1. *Gene therapy in GSD type Ia:* The goal was to evaluate AAV vectors in canine Glycogen Storage Disease.
2. *Comparison of AAV vectors for gene therapy in Glycogen Storage Ia*: The goal was to compare two vectors and to recommend the vector for use in clinical trials of gene therapy in GSD-Ia

The first grant listed was awarded in 2010 to evaluate AAV vectors in the dog model for GSD-Ia. We were pleased to publish a paper written with our veterinary fellow, Dr. Amanda Demaster, as first author. Dr. Demaster worked vigilantly with our dog colony to treat 9 GSD-Ia puppies with AAV vectors. The main conclusion from this study was that GSD-Ia could be treated very effectively with an AAV vector, although treating again with a new AAV vector was necessary from time to time to maintain control of hypoglycemia. An important consideration was whether the mother dog had formed antibodies against AAV that crossed the placenta, and AAV was less effective in that scenario.

Dr. Elizabeth Drake, veterinary fellow for the GSD Dog Colony at Duke University. Shown here with a newborn alpaca at her alma mater, Washington State College of Veterinary Medicine.

The second grant funded a comparison of two vectors in mice with GSD-Ia. The design of that study was to compare two vectors, one from Duke (reference 2 and 3, below) and one from NIH. In the end, a slight advantage for the NIH vector seemed likely. The study was suspenseful, because the vectors were "blinded" so that neither our lab nor Dr. Chou's lab at NIH knew the identity of the vector preparations that we were testing during the study. Essentially like the taste test, "Coke or Pepsi". Conducting the study was instructional in itself, and that will increase our ability to design the best studies possible in the future.

Finally, we are pursuing new ways of improving our AAV vector to more effectively cure GSD, and look forward to working with the Children's Fund in the future.

University of Florida Medical Center
David Weinstein M.D., M.M.Sc, Thomas Conlon, Ph.D.

Gene Therapy in the Canine Model of GSD Ia:

Curing GSD is the ultimate goal, and the program moved from Harvard Medical School to the University of Florida in 2005 to pursue a dream of performing gene therapy. Since 2007, 8 dogs with naturally occurring GSD have undergone successful gene therapy, and another dog will receive the treatment in the upcoming weeks. The gene therapy has turned a previously fatal disease into one that the dogs survive, and several of the dogs have already stopped treatment. Testing this year also demonstrated continued enzyme activity 4 years after the last treatment. Based upon the success of this work, an NIH grant was submitted in collaboration with GlyGenix Therapeutics, Inc (GlyGenix.com) in 2012.

These are exciting times in the gene therapy arena. A genetic form of blindness has already been treated at the University of Florida using gene therapy, and trials in humans with GSD II have commenced with early success. Gene therapy was also recently performed for another liver disease by another group using the same adeno-associated viruses (AAV) as have been used in the GSD Ia dogs. In that study, 6 people with hemophilia were treated with gene therapy, and 5 were able to stop on-going treatment. The only person that still required treatment was the person that received the lowest dose in the safety and dosing trial. With the increasing safety profile and demonstration of efficacy, we are optimistic that a trial for GSD Ia can now be approved. An application to begin human gene therapy for GSD Ia will be submitted to the FDA in early 2013, again, in collaboration with GlyGenix Therapeutics.

II. Extended Release Cornstarch:

At the 2004 meeting sponsored by The Children's Fund for GSD Research, a committee was created to look into finding an extended release cornstarch preparation. After several products failed to live up to expectations, a product was identified in 2008, and the efficacy studies were performed in Florida using support from The Children's Fund for GSD Research. The product was proved efficacious, and it was released in 2012 as a treatment for children and adults with GSD type Ia. Since being released in August 2012, over 60 people have been switched to the new product, and they are now sleeping through the night for the first time. This is the first major breakthrough in the management of GSDs since the discovery of cornstarch in the early 1970s.

On the path to 'The Cure', publications are reflective of the quality of the research and the team. Please visit **www.curegsd.org** as we update publications related to gene therapy work and other research supported by the Children's Fund for GSD Research.

The Rosen Family

Thank you Izzy Rosen and her amazing family for making our dreams of curing GSD a family priority. Izzy asked guests of her Bat Mitzvah to make donations to our foundation in lieu of gifts in honor of her cousin Jonah Feldman.

Sam & Dani Josephson

Thank you so much to Sam Josephson and his family for donating to our foundation in honor of Sam's Bar Mitzvah. We really appreciate your continued support and spreading the word to family and friends.

Kamryn Jackson

Thank you to Donna McNeil for organizing another successful raffle in honor of Kamryn Jackson!

THE ANDRUSHKO'S 9TH ANNUAL BBQ FOR GSD

Kasen Andrushko

After a wonderful turnout for our 9th BBQ for GSD research, we are excited to announce we have raised over $131,000 for GSD research!

Kasen has been selected to represent Manitoba as the Children's Hospital Champion. He will be an ambassador for all the children in the province, and help raise awareness for the hospital to raise funds for equipment, research and much more! We have already traveled to Ottawa and Florida to meet all the other Champion children throughout Canada and the Unites States. What an amazing time we had...making long lasting friendships.

We are looking forward to a new trial study that is anticipated to be starting soon at our local Children's Hospital for the new glycosade product. It will be life changing for our family. Thank you to all of our wonderful family and friends who have donated over years and who continue to support our fight for a cure for GSD. We truly appreciate you all.