One of our primary goals when forming The Children’s Fund for GSD Research over 13 years ago, was to produce enough research to attract a larger company to get involved with finding a cure. Our dream is getting closer to reality as Dimension Therapeutics, a gene therapy company based out of Boston recently announced its plans to move forward with studies focused on finding a cure for GSD1a!

Dimension Therapeutics, Inc., is a leading rare disease company advancing novel, liver-directed treatments for diverse genetic disorders. The company will be working closely with Dr. David Weinstein, one of the leading world experts in the Glycogen Storage Disease field, as well as other physicians, GSD families and patient communities to improve the lives of patients with GSD1a by advancing Gene Therapy trials. The company expects these human trials to commence in 2016.

To learn more about Dimension Therapeutics, please visit www.dimensiontx.com. You can click on Media Center to read the April 23rd Press Release announcing Dimension’s work with GSD1a.

This is an important time for everyone in the GSD community and we would not be where we are today without your help and generous contributions.

We know that we are not at the finish line yet, but we just got a whole lot closer, and it’s hard not to get excited!!

"...because every child deserves to be healthy."
Currently Supported Research

Janice Y. Chou, Ph.D., NIH

Patients with glycogen storage disease type 1a (GSD-Ia) lack the enzyme glucose-6-phosphatase-α (G6Pase-α) in their liver. All patients have problems controlling their blood glucose levels while 70-80% of patients over age 25 years also develop a long-term complication called hepatocellular adenoma (HCA). We are developing a 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 G6Pase-α gene in a novel virus-based vector, named RAAI-GPE-G6Pase. A problem in gene therapy is to get the introduced gene expressed in the right tissue at the right level to restore normal function. The key characteristic of our approach is that we use the natural human G6Pase-α gene control elements to control the G6Pase-α gene. When introduced into the GSD-Ia mice our virus delivers the gene to the liver, restores their G6Pase-α activity in the liver, and prevents the occurrence of HCA. 

The G6Pase-α gene contains the sequence of 3-letter codes that provide the genetic information to produce the G6Pase-α enzyme. Recent studies have shown that there are ways to improve the 3-letter code to make it more efficient in making the enzyme protein, named “codon optimization”. We have used this technique to optimize the expression of the G6Pase-α gene and have initiated studies to compare the efficacy of the normal and optimized versions of the G6Pase-α gene. A higher level of expression is expected to be beneficial in clinical trials. The study consisted of performing gene therapy on 2-week-old GSD-Ia mice and monitoring the correction of their disease over the next 8 to 10 weeks. When examining the amount of G6Pase-α expressed at 2 weeks after therapy, our results showed that the optimized gene produced up to 2.5-fold more G6Pase-α activity in the liver, compared to the non-optimized gene. This study is on-going but the preliminary data suggest that the optimized gene is a leading candidate for human clinical trials. 

To test how long the effect of gene therapy might last, we have done an extended study in mice. We have shown that our therapy, when given to young mice aged 2-4 weeks-old, can last for at least 70 weeks in correcting for the loss of G6Pase-α in the liver of GSD-Ia mice and preventing the occurrence of HCA. While this study shows that gene therapy can prevent HCA formation in young mice, what we still need to learn is if HCA already exists, can our gene therapy stop its progress or reverse its effects. This is important to know as it will determine who might be eligible for initial treatment when we get to the stage of conducting a clinical trial. Recent studies have shown that in a mouse where the G6Pase-α gene has been inactivated only in the liver, the mouse lives a lot longer than a mouse that has had the gene inactivated in all its tissues. In our previous experiments we have used the latter mouse, and it has not been possible to keep them alive long enough to develop HCA. Recently, the liver-specific inactivated mice have been shown to live long enough to develop HCA. So we are now using this strain of mouse to understand whether our gene therapy can slow, stop, or even cure existing HCA. Armed with this knowledge we will be ready to start the final, important studies required by FDA to allow us to initiate a clinical trial.

The GSD community lost a very special child on October 7, 2014. In Mendel’s memory, we must forge ahead, keeping him in mind and all the other GSD children and families that this disease affects. Our foundation will continue to support the most promising research for GSD-1 and as we do so, we will always have Mendel Cotlar in our memories.

Mendel Collar, a 13-year-old boy from Houston, Texas, passed away following a severe overnight hypoglycemic incident. Only three weeks prior, he had deeply inspired and impressed the many in attendance at his Bar Mitzvah celebration. Mendel was appreciated by everyone who knew him as possessing exceptional personality and character. In his encounters with people of any age, he exhibited genuine warmth, cheerfulness and enthusiasm. He would seek out and try to provide others with any kind of help they needed. He also demonstrated a sincere sense of gratitude for anything done for him and for the smallest things given to him. Mendel’s GSD was just “background noise” to him — his love of life and optimism made it irrelevant and almost unnoticeable. He loved exploring and reading, playing with his eclectus parrot, discovering new technology, and taking care of his baby brother.

Mendel is survived by a large and loving family: parents, Daniel and Eta Cotlar, brothers Nosson and Aryeh (type 1a), and sister Chani, grandparents Dr. David and Bayke Collar of Houston and Dr. Henry and Chaya Smukler of Toronto, many aunts, uncles and cousins in Houston, Toronto, Boston, North Carolina and Melbourne with whom he was unusually close.

In Memory of Mendel

In April of 2014, The Children’s Fund launched its “I am hope” campaign. We sold over 250 t-shirts all over the country. These t-shirts symbolize our hope that we will indeed find a cure for GSD soon! We have limited sizes left, but we are still selling t-shirts on our website for $35. 100% of the sales price goes directly to fund promising research. We all have hopes and dreams…Hope for world peace, hope for equality amongst all people, hope for everlasting love…Our greatest hope and dream is to cure GSD. Whatever your cause, we hope your dreams come true. Thank you for supporting ours! Please visit www.cureGSD.org to purchase a shirt.
The Children's Fund for Glycogen Storage Disease Research was founded in April 2002 by families of children with GSD1a who wanted to make a difference. At that time there was very little support for someone with GSD1a and no research being done. Since then, our foundation has grown as more families have joined together, all with the common goal of curing GSD. We are now the world’s leading GSD1 organization, dedicated to funding research so that children born with GSD1 will benefit from early detection, treatment and an eventual cure.

In an effort to minimize expenses and maximize the funds available for research, The Children's Fund for GSD Research is managed solely by volunteers. Our Board of Directors is made up of parents and relatives of children with GSD and all fundraising is done on a 100% volunteer basis. Each grant submitted is reviewed by a Scientific Advisory Board made up of leading physicians and scientists with extensive knowledge of Glycogen Storage Disease. Their reports are submitted to the Board of Directors for review to insure the most beneficial use of our funds.

Since 2002, we have raised over $6.5M and granted almost all of this to support over 40 research studies.

The results from these studies were instrumental in curing GSD1a in mice and dogs; FDA approval of Glycosade (superstarch); adding GSD to the genetic testing panel; preventing and treating adenomas; and securing a large pharmaceutical company to fund human gene therapy trials expected to commence in 2016.

Since GSD is an orphan disease, The Children's Fund receives no federal funding. All money raised is through fundraisers organized by families and friends of the foundation. Our Board of Directors hosts two events each year, a Super Bowl raffle and an Annual GSD Gala in South Florida. These, along with the many events that take place around the country throughout the year (golf/tennis/basketball tournaments, cocktail parties, BBQs, wine tastings, road races, concerts, garage sales, etc.) allow us to fund the research that will eventually lead to a cure for GSD1. We encourage you to get involved.

To learn more about The Children’s Fund for Glycogen Storage Disease Research and how you can help, please visit us at curegds.org.
In The News

A Night at the Derby
The 8th Annual GSD Gala, A Night at the Derby, took place on April 26, 2014 in Weston, Florida. Over 400 people were in attendance and it was truly an inspiring night. Guests wore their best derby attire and got excited over three horse races throughout the evening. The evening featured mint juleps, southern fare, an exciting live auction and outstanding silent auction that made this year’s event the most successful to date. We were honored to have Dr. Weinstein in attendance, and the attendees were excited to hear him speak about his ongoing research and progress towards a cure. It was a moving, meaningful and inspiring evening and we are pleased that the money raised is being used to support research bringing us closer towards human clinical trials with the FDA.

Zack & Jake’s Basketball Tournament for GSD
Our 2nd basketball tournament to raise money for GSD had an amazing turnout. It was held on June 7th, 2014, and we were fortunate to have a lot of support from Zack & Jake’s friends. They all came out to play in the 8th grade basketball tournament in support of GSD. We had a great deal of support from many local stores and vendors in Westchester who gave us food for the night and items to raffle off in the auction. It was a fun filled night with great basketball, a silent auction and a raffle auction. We reached our goal and raised $12,000 for GSD funding and research. We are grateful for all the support we have received!

Thank You to Ed and Corinne Rennie
for making an extremely generous donation to our foundation in honor of Justin Bagin

Christa’s 40th Birthday
Happy Birthday to Christa Gurka and a big “thank you” from The Children’s Fund for asking your party guests to make donations to our foundation in lieu of birthday gifts. We are so lucky to have friends and supporters like you!

A Thank You From Jonah Feldman
Thank you to everyone who contributed to The Children’s Fund for GSD Research for my Bar Mitzvah. We raised over $40,000 on November 8, 2014! My family and I really appreciate all of the love and support you have given us over the past 13 years. We couldn’t have done this without you.

Love,
Jonah Feldman

Super Bowl XLIX
Congratulations to Kelly Moran and her husband Steve of Greenwood, CO, who were the Super Bowl raffle winners. There were 688 tickets sold, and with matching gifts, over $70,000 was raised.

Thank you to the Reed family for running this raffle in honor of Anna.

Carla Rides for GSD
On May 3, 2015, Carla, friend of the Reed family, embarked on a 5-Boro Bike Tour to benefit The Children’s Fund for GSD Research.

On April 25, 2015, The Children’s Fund for GSD Research hosted its 9th annual gala at the Design Centers of the Americas (DCOTA building) in Dania Beach, Florida. This year’s theme, “Unmask the Magic” featured a world renowned magician who wowed the crowd throughout the evening with his close up magic and mentalism. Close to 400 attendees donned their most elaborate masks in support of The Children’s Fund’s tireless dedication towards funding promising research in an effort to cure GSD1a. Most notably, this year’s event was the most successful event to date, thanks to the many sponsors and guests who return to support our cause year after year, and to the new faces that are passionate about what The Children’s Fund is all about! We excitedly announced Dimension Therapeutics’ involvement in helping with human clinical trials, and all who attended were inspired by the progress that a foundation of our size, with limited reach, has achieved. “Unmask the Magic”, co-hosted by Lisa Hodes and Meredith Gussin, proved to be a memorable, exciting and inspiring evening! Evenings such as this infuse more passion and dedication in all of us at The Children’s Fund and we are beyond thankful to all who attended and helped make the night such a success!