

The Children's  
Medical Research  
Foundation, Inc.®

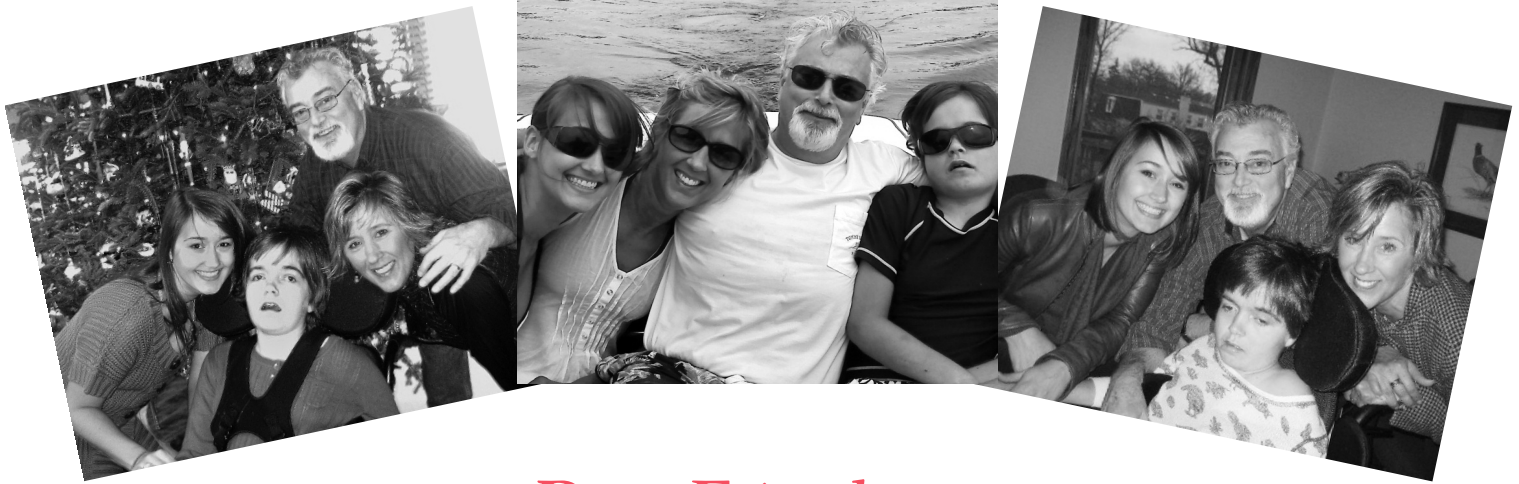


# KirbyGram

*The latest news on Kirby Wilson and friends and the search for a cure for Sanfilippo Syndrome*

**December 2011**

*Our goal is to create awareness of Sanfilippo Syndrome and other neuro-genetic disorders, fund medical research and find a cure.*



*Dear Friends,*

*As Kirby's story continues to unfold, there are times Brad and I sit and allow ourselves to wonder. Does she still know our voice, our face or the depth of love we feel when we see hers? Does she know it is we who hold her close for comfort and to feel her strength and tenacity from within? Does she remember her love of song and dance and fun -- when it was she wondering, "Are you happy?" The happiness of those around her was of the utmost importance.*

*When Brad and I think of happiness, we think of our children and family. But we also think of you and the happiness you have brought to our lives and the accomplishments you made possible for The Foundation over the past 17 years. As we stand on the threshold of a cure, we reflect on the success story you are helping to create. The story of hope for thousands of children. The story of honor and happiness for Kirby. Brad and I are blessed to be Kirby's parents and honored to call you our friends. Yes, Kirby, we are happy.*

*This holiday season, our wish for your families is happiness, the same happiness Kirby brings to our lives each day. We ask that you think of Kirby, and in her honor, continue to support The Children's Medical Research Foundation as it strides ever closer to its mission of a cure.*

*May the blessings of the season be yours, now and always.*

*– Sue and Brad Wilson*



# *Glad Tidings From The National MPS Society*



The National MPS Society chose Kirby as one of this year's Standing Ovation Award recipients. This award reads "In honor of your resilience, courage, tenacity and passion for life while living with the many challenges of having MPS or a related disease." It was a wonderful surprise for the Wilson family. Sue comments, "This type of recognition is not something children like Kirby receive very often, so it was a pretty great day for us all."

The National MPS Society's mission is to find cures for MPS and related diseases. The organization provides hope and support for affected individuals and their families through research, advocacy and awareness of these devastating diseases.

## *Fundraising News*



### **"Fore" Kirby**

June 3rd was the date of the Sixteenth Annual "Fore" Kirby Golf Fun Raiser at Ruffled Feathers Golf Club in Lemont. This year's event had just 16 participants, yet raised more than \$33,000 "fore" Kirby!

Sue comments, "This is always a fun event for Brad and me as it enables us to relax with friends while raising money each year. We are very fortunate to have these people in our lives,

always willing to help us as a family or do whatever it takes for The Foundation to succeed."

The Foundation is actively seeking new participants for this unique golf outing. The event is held each year on one of the first Fridays of summer. Participants are given pledge cards and are asked to secure pledges from friends and associates for each of the 18 holes of golf to be played.

Upon completion of the round, score cards are collected from

each player. The Foundation then contacts all of those who have pledged with the results of their player's round and the total amount due. Participants also are asked to pay for their round of golf, which means that 100% of the donations go directly to The Foundation! Cocktails and dinner are served immediately following at the Wilsons' home.

Interested in joining the fun? Please contact Sue at (708) 784-0631 or [curekirby@sbcglobal.net](mailto:curekirby@sbcglobal.net) to learn more.

### **Junior Board Makes Its Mark**

The Foundation's Junior Board hosted its first fundraiser, "Cruisin' for a Cure," on September 16th in Chicago. Maggie Wilson and fellow Board members Joe Avram, Kelly Brummet, Ryan Brummet,

*(Continued on Page 3)*

# Fundraising News



**Junior Board member Kelly Brummet with her mom, Kathi, and Maureen Blaber.**

Bob Karpiak, Kristin Pipal and Molly Phillips organized this two-hour evening cruise on Lake Michigan, complete with music, cocktails, food, raffle and an after-party. The event raised more than \$1,100 with 51 guests. Brad and Sue were delighted to attend the event and privileged to see, firsthand, this Board's enthusiasm toward being a part of The Foundation's success.

## Maas Gets Personal

Marty Maas of Maas Auto in LaGrange took it upon himself to fundraise toward a "cure for that beautiful little girl." That beautiful little girl being Kirby Wilson. On November 6th, with help from Bronko Palikuca of Topaz Café in Burr Ridge and a group of its employees, Marty hosted his first "Take A Stand Against Sanfilippo" luncheon. Over 80 of his loyal customers attended the event, with Topaz employees not only choosing to come in on a Sunday to work, but also donating all gratuities.

The luncheon raised more than \$7,400. Sue comments, "Brad and I live in a wonderful, caring community of people who have stood by our side and supported our mission for over 17 years, with each passing year attesting to the depth of the compassion."

## The Foundation Gives Thanks . . .

To **Margaret Dawe, Nicholas Megofna, Mark Leavitt and Donna-Logan Gabel, Raymond Donato, Ronald Odrobinak, William Leonard and Renata Fetzer**, who designated The Foundation as their charity of choice in their employers' United Way campaigns. Thanks for uniting for Kirby!

To **The Marmon Group** for its matching contribution, doubling employee **James Angus's** donation to The Foundation. It's a great match for Kirby!

To **Barbara Burkhardt**, who donated in memory of **Brian Noel**.

To the many friends and family members of **Ellie Rounce** who honored her life with memorials to The Foundation. Ellie was a longtime friend of the Wilsons, and one of her last wishes was to raise a million dollars for Kirby. She was a beautiful, kindhearted woman who will be missed by many.

To the many contributors who used the donation envelopes as an opportunity to give to The Foundation. Donations from our June newsletter totaled \$905.

To **Kirschbaum's Bakery and Casey's Market**, both from Kirby's hometown of Western Springs, for their continuous fundraising efforts using cash jars. And to the people of the community who fill them and have helped to raise more than \$650 to date this year, your "hometown girl" thanks you from the bottom of her little heart.

## And To The Families Working Together For The Cure . . .

Brooke and Ashleigh Kidwell of Louisville, KY were honored with birthday donations from Grandma and Grandpa Kidwell, Mr. and Mrs. Larry Sinn and Judy Bible. Now that's a celebration!



**Les Sheaffer with his girl Brittany.**

**Les and Teri Sheaffer** of Kempton, PA, honored their daughter **Brittany** with a donation to The Foundation.

**David and Barbara Trocheck** of Diamond, OH, passed on the proceeds from the Third Annual Hiram Golf Outing, which honored their daughter **Carley** and our campaign for the cure.

*(Continued on Page 4)*

# Fundraising News

Carley's physical therapist, **Mandy Zukowski**, and her husband, **Bruce**, were hosts of the event.



**Carley Trocheck**

## News From Connecticut

### *A Note From Rhianna's Mom – Cynthia Logan*

Another year has flown by, and we feel truly blessed by the wonderful support we have received this year after our plea last fall. After 16 plus years it is hard to find a different way to say thank you or to describe the gratitude in our hearts for those who have worked so hard for Rhianna, for a cure. Rhianna continues to stay connected and shares those very precious smiles. Please picture one of those smiles specially wrapped and sent first class to each of you who continues to give us hope.

Every day I wake up next to her and wish I could spend the day seeking the funds to push the research over the top, and there she is with so many needs. It is very exciting to see the new possibilities that exist, such as the Pepsi Challenge, the Chase Foundation and a superb National MPS Society. We can all sit in the comfort of our homes and just click on our computers and have such great potential for change.

I thank our family and friends who have supported these opportunities, as well. Our hope can stay strong knowing that technology is our new fundraising friend.

I cannot close without a special thanks to Sue and Brad Wilson and the extraordinary group of researchers that they have funded. Without their 17 years of outstanding dedication and commitment, we all would not be funding breakthroughs. We would still be trying to find the responsible gene. I recall being told at the time of Rhianna's diagnosis in 1995 that we would not see even a treatment for any of the MPS disorders in our lifetime. A cure was out of reach. Look how far we have come. The groundwork that supports the current efforts is really quite remarkable. May the Wilsons, the researchers and the new supports remain strong so our hope can become a reality.



**Cynthia, Rhianna and Gene Logan with the Theriaults - Alan, Michael, Marissa and Donna.**

Longtime friends and supporters **Donna and Alan Theriault** organized their Second Cutting for a Cure cut-a-thon/spa-a-thon. For ten years they helped to raise

funds with their Rockin' for Rhianna fundraiser, inspired by Rhianna's courageous battle with Sanfilippo. This year's event, held September 18th, raised more than \$5,700. We thank the Theriaults for their tireless efforts on behalf of Rhianna.

Our thanks go to **Tabitha and Justin Manafort** for once again giving the proceeds of the **Manafort Family Foundation's** annual golf outing to The Foundation.

**The Roberts family** continued its support of our mission of a cure with a donation in honor of Rhianna through **Clinton S. Roberts Foundation**.

**Dennis and Laurel Colgan of Picture Fame** continued their support of "Rhianna's Hope" with a donation to The Foundation.

A donation honoring Rhianna also was received from **John St. Pierre of Saints Restaurant**.

**An Artisan's Marketplace** passed on \$100, which was the money collected from "Rhianna's cash jar."

**Susana Tesfaye, Joe and Lynn Fuda and Donna and Alan Theriault** celebrated Rhianna's 19th birthday with a donation. Happy Birthday, Rhianna!

And last but certainly not least, a donation was received from Rhianna's grandparents, **James and Martha Couture** in recognition of Rhianna's graduation from high school and those who worked so very hard to help her reach this momentous occasion. Our congratulations to the graduate!

# Research Update

**Update from the Laboratory  
of Dr. Elizabeth F. Neufeld,  
Department of Biological  
Chemistry at UCLA,  
November, 2011**

Our studies of the Sanfilippo syndrome (MPS III) continue along two lines – what is wrong in the brain and how can we treat it? We use the MPS IIIB mouse model for these studies. The primary defect in MPS IIIB is a mutation in the gene (*Naglu*) encoding  $\alpha$ -N acetylglucosaminidase, one of the lysosomal enzymes needed for the breakdown of heparan sulfate. As a result, there is a deficiency of  $\alpha$ -N-acetylglucosaminidase, and heparan sulfate accumulates in lysosomes. But in addition, there is a small area of the brain (the medial entorhinal cortex or MEC) where a number of apparently unrelated substances accumulate; one of these also accumulates in another small area, the dentate gyrus. The medial entorhinal cortex and the dentate gyrus, which are connected to each other, are involved in learning and memory. While our primary study system is the mouse model of MPS IIIB, we have found similar pathology in the brain of the mouse model of MPS IIIA, which is lacking another lysosomal enzyme of heparan sulfate degradation.

The diverse substances that accumulate in the MEC include (among others) proteins modified by ubiquitin, proteins involved in autophagy, and proteins that tend to aggregate. The latter include lysozyme, hyperphosphorylated tau (Ptau) and amyloid beta; the

last two are known for their involvement in Alzheimer disease. Most importantly, we have found accumulation of glypican, a heparan sulfate proteoglycan (HSPG) molecule in which the heparan sulfate is attached to a protein backbone. HSPG is the form in which heparan sulfate is made by the cell and in which it functions. After fulfilling its function, the HSPG normally enters lysosomes, where the heparan sulfate part is separated from the protein in order to be broken down. In MPS III, the heparan sulfate cannot be broken down and therefore accumulates inside lysosomes.

Our hypothesis is that neurons can tolerate a certain amount of lysosomal storage of heparan sulfate. But in MEC, the lysosomes are so loaded that they have trouble receiving any more HSPG, which causes a back-up in the cellular trafficking of this substance. The HSPG outside of lysosomes can interact with amyloid beta and Ptau and cause these to form aggregates that are hard to break down as well as toxic to the neuron. We plan to test this hypothesis by various biochemical and molecular methods, as it has implications for developing treatment.

Each step in the cascade of events, from synthesis of HSPG to aggregation of Ptau, can be considered a drug target. We previously showed that the drug lithium chloride could reduce Ptau aggregates in the dentate

gyrus. Lithium inhibits an enzyme which puts the phosphate on tau in order to make Ptau. We are now working with a compound that helps (indirectly) to remove the phosphate from Ptau, and will test it in combination with lithium chloride. Another step that may be easy to affect is autophagy. The ideal treatment would be to make HSPG with shorter branches of heparan sulfate so that less would accumulate in lysosomes, a treatment called substrate reduction therapy. We note that by our hypothesis, we would not need to reduce lysosomal storage of heparan sulfate to the normal level, but only to a level that would make it tolerable to MEC neurons by preventing the trafficking back-up of HSPG and the additional accumulations.

*The studies mentioned above have been published: Ohmi K, Zhao HZ and Neufeld EF, PLoS One. 2011;6(11):e27461.*

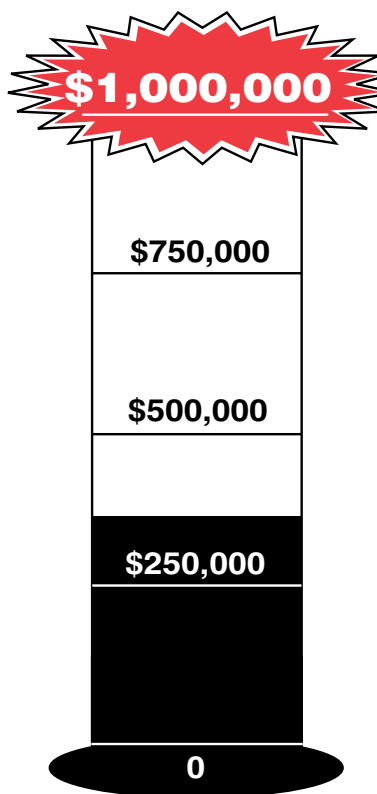


(Continued on Page 6)

## Update On Funding Studies

Over the past year, we have spoken to you about our campaign to raise \$1,000,000 to support Dr. Haiyan Fu of The Research Institute at Nationwide Children's Hospital in Columbus, OH, and the studies required for FDA approval of human trial.

We are happy to report that through your support and the efforts of other families and foundations, funds have been raised and grants issued to Dr. Fu and The Research Institute at Nationwide that now total \$275,000. And, with each passing month, more and more families are joining forces to make the \$1 million a reality.



It is The Children's Medical Research Foundation's goal to continue to support these studies, as well as other research offering hope of a lifesaving treatment.

We are asking you to visit our website at [www.curekirby.org](http://www.curekirby.org) and go to its Blog. Click on "A Cure is Within Our Reach." It will allow you to be one of the first to watch our You Tube video. Donate what you can, whether it be \$1, \$5 or \$100. Then tell friends and ask them to do the same. Help us to make a cure for Sanfilippo the next You Tube sensation!

Want to get involved? Please read on and take a look at the ways you can help us achieve our mission of a cure with these fundraising ideas.

## Fundraising Opportunities

### Have Fun With This Office "Fun" Raiser – Kirby Dares You!

Here's a fundraising idea that can be challenging and fun for everyone in your office. Are you willing to take a dare for Kirby? Here's how it works. Your company pledges a total amount it would be willing to donate to The Foundation. Then each employee willing to take on a dare chooses the dare and the amount to be donated if he or she follows through. There can be a list of suggested dares with donation amounts, or you can leave it up to the employees to get creative and have some fun. Do a dance or perform a song on the street, or for the office, wear heels for a day (that would be a guy thing), get a Mohawk, do cartwheels down a hall, eat or drink a mystery

concoction...all for fun and a great cause.

### A Match For Kirby

Does your company have a matching gift program? It could double your support of The Foundation.

### United Way Can Be For Kirby, Too

Does your company have United Way pledges at your workplace? Although we are not a United Way member, you can designate The Children's Medical Research Foundation as your recipient, and the funds will be forwarded to us through the United Way Campaign! Simply give your local United Way agency The Foundation name, address and our Federal ID #36-4033667.

### Celebrate, And Make It For Kirby

Is there a special birthday coming up for a family member or friend? Are you looking for an alternative to the typical "over the hill" gift? Be different. In lieu of gifts, donate to The Children's Medical Research Foundation. Kirby always loves a party!

### A Gift Like No Other

This holiday season give clients a donation to The Children's Medical Research Foundation in their name. It's a gift that won't gather dust and goes far beyond any other.



# *The Sweetheart Dinner Dance*

**February 10, 2012 • Four Seasons Hotel Chicago**

*An elegant setting at one of the world's finest hotels*

The 17th Annual Sweetheart Dinner Dance will be held Friday, February 10, 2012, in the Grand Ballroom of the Four Seasons Hotel Chicago.

This gracious venue is set high above the Magnificent Mile, where guests will be welcomed with a champagne cocktail reception.

Executive Chef Kevin Hickey will serve a four-course dinner in contemporary American style enhanced by the great bounty of the Midwest.

We will be treated to the magical sounds of the Michael Lerich Orchestra, which has entertained us for the past 16 years! And silent auction packages promise to tempt sports, travel, dining, spa and shopping enthusiasts.

Reservations are \$250 per person or \$2,500 for a table of ten and must be made in advance. Invitations will be mailed in December, or you can reserve your place now online at [www.curekirby.org](http://www.curekirby.org). Please plan to join us by marking your calendars now.

♥ **We need your help to make it a success** ♥

This dinner dance is the primary fundraising event of the year, and we're asking you to help ensure its success.

We have designated five sponsorship levels for the Sweetheart Dinner Dance, as listed below. In appreciation of your sponsorship, you will receive prominent event recognition. We ask that you indicate your wishes on the following Reply Form and return it to The Foundation or visit us online at [www.curekirby.org](http://www.curekirby.org). Please contact Sue Wilson at (708) 784-0631 or [curekirby@sbcglobal.net](mailto:curekirby@sbcglobal.net) with any questions.



## **The 17th Annual Sweetheart Dinner Dance**

### **~ SPONSORSHIP REPLY FORM ~**

***Sponsorship:***

- |                                   |          |                                     |         |
|-----------------------------------|----------|-------------------------------------|---------|
| <input type="checkbox"/> Diamond  | \$10,000 | <input type="checkbox"/> Silver     | \$1,000 |
| <input type="checkbox"/> Platinum | \$5,000  | <input type="checkbox"/> Sweetheart | \$500   |
| <input type="checkbox"/> Gold     | \$3,000  |                                     |         |

Name as you want it to appear (*Please print*): \_\_\_\_\_

☐ Check Enclosed

Please charge my (*Check one*):    Visa/MC \_\_\_\_\_    American Express \_\_\_\_\_    Discover \_\_\_\_\_

Cardholder Name (*Please print*): \_\_\_\_\_

Account Number \_\_\_\_\_

Expiration Date \_\_\_\_\_    Signature \_\_\_\_\_

Please include your phone number \_\_\_\_\_

Please return this form to: **The Children's Medical Research Foundation, Inc., P. O. Box 70,  
Western Springs, IL 60558, fax to (708) 784-1978 or call (708) 784-0631.**



PRESORTED  
FIRST-CLASS MAIL  
U.S. POSTAGE  
PAID  
MAILED FROM  
ZIP CODE 60477  
PERMIT NO. 222

**The Children's  
Medical Research  
Foundation, Inc.®**

P.O. Box 70  
Western Springs, IL 60558

**KirbyGram**



# Save The Date

**Friday, February 10, 2012**

**The Sweetheart Dinner Dance Makes  
a Romantic Holiday Gift**



This holiday season, give your sweetheart a gift of good cheer – a romantic evening  
at the Sweetheart Dinner Dance.

You'll avoid last minute shopping in crowded stores when you call  
(708) 784-0631 now for reservations.

Then enjoy February 10th in taffeta and tux, sipping champagne, dining in the  
Grand Ballroom of the Four Seasons Hotel Chicago and dancing to the music of the  
Michael Lerich Orchestra. Cheers!



*Copyright 2011 The Children's Medical Research Foundation, Inc. All rights reserved.  
Reproduction prohibited without the express written consent of The Children's Medical Research Foundation, Inc.*