



KirbyGram

December 2009

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

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

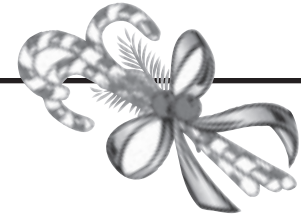
Kirby Update

Kirby is enjoying her fourth year at Lyons Township High School with Jackie Gay, the new classroom teacher. Jackie is working hard to ensure Kirby does not miss out on any activities because of her new medication schedule. When possible, Jackie herself takes Kirby on community outings where Kirby is treated to a little one-on-one with the teacher. This is Jackie's first teaching job, and she brings fresh ideas to the room where staff members continue to provide an outstanding environment for Kirby and her fellow students.



The happy school girl.

Sue explains, "As I have said before, Kirby's room is filled with people who believe in their students, celebrate their abilities, nurture their strengths and are always looking for innovative ideas to help the students to confront their challenges with the dignity they so richly deserve." Sue continues, "Jackie embraces these qualities with her own cheerful enthusiasm. We remain grateful to the entire C115 staff."



Dear Friends,

When Brad and I formed this Foundation, it was because of our four-year-old daughter, Kirby, the disease she was afflicted with and the hope that Sanfilippo and its devastating progression could not possibly manifest itself within her -- our beautiful bundle of joy. The solution seemed simple. Raise funds to enable researchers to advance and expand their work to find a cure. We chose to fight this disease.

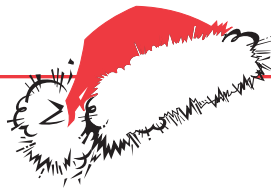
Now, 15 years later, our mission continues still because of our tenacious daughter. Kirby's resilience is inspiring. The simplest task, as in bringing her hand to her face, is a struggle for control, but her perseverance shines through as she raises her hand and it touches her face-- defying Sanfilippo. Her fight continues. As do the advances scientists are attaining. Advances that are realized because of your enduring commitment to our Foundation and its mission of a cure. We are grateful that you too continue.

This holiday season, Brad and I wish for your families and you the same joy 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, its mission and the researchers who are working so diligently to find a cure.

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

Brad and Sue Wilson

Fundraising News



"Fore" Kirby

The Fourteenth Annual "Fore" Kirby Golf Fun Raiser was held May 29th at Ruffled Feathers Golf Club in Lemont. This year, 16 participants raised more than \$22,000 "fore" Kirby! Nanci Makris and The Flame of Countryside once again helped to provide a delicious dinner buffet at the Wilsons' home after the round of golf.

Sue comments, "This year's group was one of the smallest yet, but still raised an incredible amount of money for the Foundation. Brad and I are very fortunate to have such caring people willing to do whatever is asked of them to help us raise funds and awareness. This event reminds us of how privileged we are to call these people our friends."

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, and 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 to learn more.

The Foundation Gives Thanks....

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

To **Bill and Karen Rajki** for their donation in memory of Robert Smith.

To the **Fred Sammons Family Foundation** for its most generous support of our mission.

To the many contributors who used the donation envelopes as an opportunity to give to the Foundation. Donations from our June newsletter totaled \$1,360.

To **Kirschbaum's Bakery** and **Casey's Market**, all 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 \$400 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....

To Grandma and Grandpa Kidwell for their donations in celebration of Brooke and Ashleigh Kidwell.

To Barry and Nancy Boyer and Drs. Glenn Bloiso and Margaret Crabtree for their donations in honor of Sydney and Hunter Moff.



Brooke Kidwell

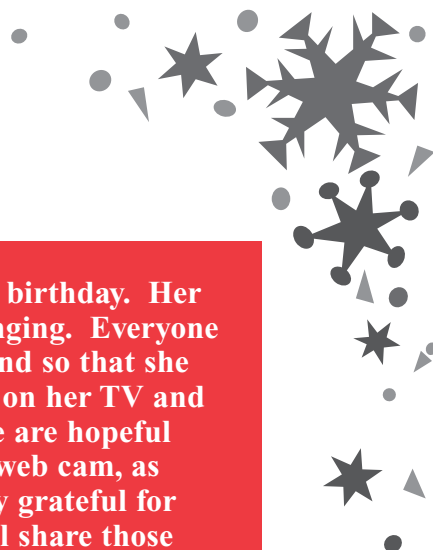
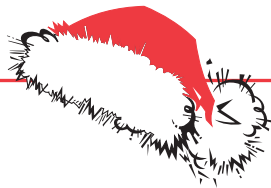


Ashleigh Kidwell



Fundraising News

A Personal Note from Rhianna's Mom



On October 15th, our Rhianna celebrated her 17th birthday. Her favorite part of birthdays is still candles and the singing. Everyone was quite generous in contributing to her laptop fund so that she can view her computer programs in a large format on her TV and visit some of her favorite people via a web cam. We are hopeful that her old dolphin friends will be first up on the web cam, as they evoke the greatest smiles. Gene and I are truly grateful for Rhianna's continued strength that allows her to still share those very precious smiles.

Sadly, within days and weeks of Rhianna's celebration, the harsh reality of MPS struck close to home with the loss of two very special friends, Paul Adams and Diana Rodrigues. It was an honor to know both of them. Paul shared a legacy of beautiful artwork, bright, colorful and full of meaning, much like Paul himself, and it is clear that Diana's shining spirit lives on in her siblings Peter and Emma. I thank their families for supporting our fundraising efforts over many years, and I am truly grateful for the strength and courage they have shared and continue to share with us.

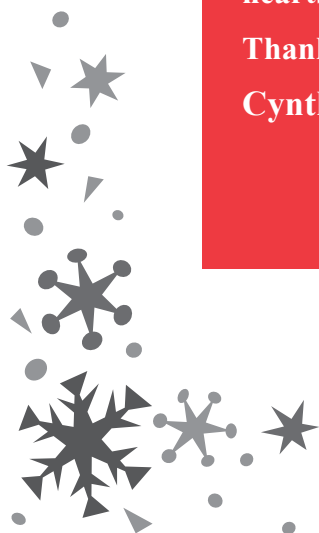
I offer this news to all those who support Rhianna's Hope and The Children's Medical Research Foundation not to cause sadness, but rather to validate the importance of all that has been done and continues to be done to fund medical research. The significance of the Foundation's efforts is grand, as it currently offers hope, generates progress toward a cure and will one day be the force that alters the course for all children and their families affected by Sanfilippo. Without each contribution, this would not be possible. Words seem inadequate to express our gratitude to each of you who give so willingly after 14 years and to the Wilsons who continue with diligence. The hope that is generated warms our hearts and lifts our spirits, and I believe it keeps Rhianna smiling.

Thank you so very much.

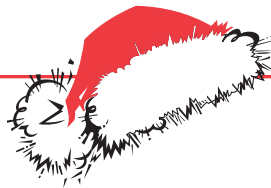
Cynthia Logan



Michael Theriault hitting the street for Rhianna.



Fundraising News



News From Connecticut

Donna Theriault's hair salon, Studio 466, was all about "Rhianna's Hope" on October 4th as this year's host of "Cutting for a Cure." Donna, Debra Dawson, Linda Casorio and Studio 466 employees joined forces to provide food and salon services that day and to solicit donations for a Sponsorship Board and raffle, as well. Over \$5,800 was raised for the Foundation by this group, that just won't quit when it comes to its own hometown sweetheart, Rhianna Logan.

Speaking of not quitting, An Artisan's Marketplace and Cassille's Restaurant seem to us to also be "regulars" when it comes to their fundraising for Rhianna. An Artisan's Marketplace, which is busy preparing for its annual fundraiser, "Stars for Hope,"

always has a collection jar on its counter, as does Cassille's, whose patrons are great at keeping it full.

We also give thanks to ...

The Manafort Family for once again including the Foundation as one of the benefactors of its annual golf outing.

Len and Gail Roberts of The Clinton S. Roberts Foundation for honoring Rhianna's 17th birthday with their continued support and hope for a cure.

Rodney and Janice Reynolds for honoring Cynthia Logan's 50th birthday with a donation to the Foundation.

Dennis at Picture Fame and Joseph and Denise Carabetta for their donations in honor of Rhianna.



Scott Haney joins Donna, Silvana, Marissa and Joey as he gets his cut.

Research Update

Update from the Laboratory of Dr. Elizabeth F. Neufeld, Department of Biological Chemistry at UCLA, November, 2009

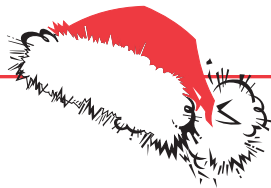
Why is the brain so severely affected in patients with Sanfilippo Syndrome (MPS III)? To answer this question, we generated a mouse model of MPS IIIB ten years ago, with funding from The Children's Medical Research Foundation and the National Institutes of Health. We are beginning to see the answer. It is more complicated than we had anticipated, but may give us leads to better strategies to develop treatment.

The primary defect in MPS III B is mutation in the gene, NAGLU, which leads to a deficiency of the enzyme alpha-N-acetylglucosaminidase and a storage of undigested heparan sulfate or its fragments in lysosomes. The original understanding was that the lysosomal storage interfered with the function of the cell by taking up a lot of space. However, that is not the case for the Sanfilippo Syndrome. The storage of heparan sulfate in liver lysosomes does not cause liver disease. But the storage of much smaller quantities of heparan sulfate in the brain causes very serious disease. This has led to a search for secondary defects in

(Continued on Page 5)



Research Update



(Continued from Page 4)

the brain. From data gathered several years ago, we had come to appreciate that many secondary accumulations, such as gangliosides, cholesterol, ubiquitin and other substances, occurred in a very small area of the mouse brain called the Medial Entorhinal Cortex (MEC). To see what was so unique about this area, we dissected neurons from the MEC of MPS IIIB mice and of unaffected control mice, as well as from a neighboring area (the Lateral Entorhinal Cortex or LEC) that does not show these secondary accumulations. Analyzing gene expression in these neurons gave us a lead about a protein, lysozyme, which was elevated in MEC neurons of the MPS IIIB mouse brains but not LEC neurons. Lysozyme (a protein associated with inflammation, which had not been previously reported in neurons) is a protein which is thought to aggregate very easily, and we postulated that lysozyme accumulated in neurons because it aggregated and then could not be broken down. There was a report in the literature that aggregated lysozyme was toxic to neurons and could lead to the accumulation of hyper-phosphorylated tau (P-tau, a protein that accumulates in the brains of patients with Alzheimer's disease). We therefore searched for the

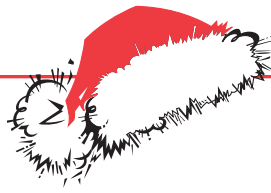
presence of P-tau in the MPS IIIB mouse brain. Indeed, we found P-tau in the MEC of MPS IIIB mice as well as in the dentate gyrus (an area to which MEC is connected). We also found an elevation in MEC of an activated form of the enzyme GSK3b, which is thought to be responsible for the formation of P-tau. GSK3b could be a target for therapy because there are many compounds that reduce its activity, the best known of which is lithium. We put some mice on a diet supplemented with lithium salts, but our pilot experiment did not show any improvement in the pathology. Perhaps we did not use a sufficient dose or start early enough; we will therefore repeat the study under somewhat different conditions. Finally, we have just obtained evidence for aggregates of Ab, another feature of Alzheimer's disease in the MEC of MPS IIIB mice.

Our current concept of the pathogenesis of MPS IIIB is that the MEC is an area of the brain in which certain proteins are prone to aggregate – lysozyme, P-tau and Ab. Whatever it is in the MEC environment that predisposes to this aggregation, it eventually must connect to the primary defect – an inability to break down heparan sulfate. We can see that there may be many ways to provide treatment, each with its own advantages

and disadvantages. Clearly, gene therapy would be ideal, but it is not yet ready for human use, although clinical trials are appearing on the horizon. Enzyme replacement would also be good, provided there were methods for delivering enzyme across the blood-brain barrier. But, unfortunately, these therapeutic methods are not yet available. We can also focus on the secondary defects. These could be inhibitors of GSK3b or they could be compounds that interfere with protein aggregation. Because protein aggregation is thought to be linked to dementia, such drugs might improve the quality of life for patients and their families. Finally, if we understood what it is about the MEC area that facilitates the aggregation of proteins, we could target that aspect of the disease.

Reference: Ohmi K, Kudo L, Ryazantsev S, Zhao HZ, Karsten SL, Neufeld EF: Sanfilippo Syndrome Type B, a lysosomal storage disease, is also a tauopathy. *Proceedings of the National Academy of Science* 106: 8332-8337, 2009.





Fundraising Opportunities

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.

Give Kirby Security

Tired of taxes? The Foundation now has a brokerage account available that allows you to donate appreciated securities. Why pay tax on the gains when you can realize a charitable deduction of the full market value of your stocks . . . and it's for Kirby, too! Contact Sue Wilson at (708) 784-0631 to learn more.

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 Match for Kirby

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

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.

Go Kasual For Kirby

Tired of wearing those heels, hose or ties five days a week? Why not suggest a "Go Kasual For Kirby Day" to

your employer? It's a great way to give a "relaxing feel" to a workday and raise funds for the Foundation. Simply send a memo to co-workers explaining the day and set the "fee" to participate.

Still in school but want to help? How about a "Hat Day" done the same way!

It's fun for the kids and a great way for them to participate in a good cause. No "fee," just leave an amount up to them.



THE SWEETHEART DINNER DANCE

FEBRUARY 5, 2010 • FOUR SEASON HOTEL CHICAGO

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

The 15th annual Sweetheart Dinner Dance will be held Friday, February 5, 2010, in the Grand Ballroom of the Four Seasons Hotel Chicago.

Our gracious new 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 14 years! And silent auction packages promise to tempt sports, travel, dining, spa and shopping enthusiasts.

Reservations are \$225 per person or \$2,250 for a table of ten and must be made in advance. Invitations will be mailed in December. 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. Please contact Sue Wilson at (708)784-0631 with any questions.

The 15th Annual Sweetheart Dinner Dance

~ SPONSORSHIP REPLY FORM ~

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

Name as you want it to appear (Please print): _____

☐ Check Enclosed

Please charge my (check one) Visa/MC _____ American Express _____

Cardholder Name (print) _____

Account Number _____

Expiration Date _____ Signature _____

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.*

Please include your phone number: _____





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

February 5, 2010

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 5th 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!