



KirbyGram

June 2004

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

What is Sanfilippo Syndrome?

Sanfilippo Syndrome is one of seven Mucopolysaccharide (MPS) disorders. There are four different enzyme deficiencies that cause Sanfilippo. The Sanfilippo disorders are described as type A, B, C, or D. There is very little difference between the four types, though there have been a few very mild cases of the B form reported where the children have remained relatively healthy into early adult life.

Children with Sanfilippo are missing an essential enzyme that breaks down a complex body sugar called heparan sulfate. This sugar slowly builds in the brain, stopping normal development and causing hyperactivity, sleep disorders, loss of speech, dementia and typically death before adulthood. There is no cure yet.

While Sanfilippo occurs once in 24,000 births, successful research into the disease could apply directly to many of 5,000 other genetic disorders.

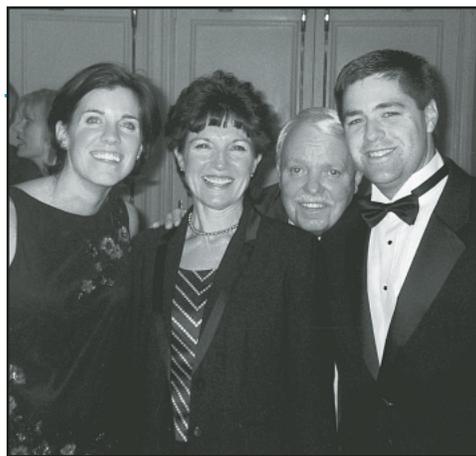
KIRBY UPDATE

Once again, the Wilsons traveled to Florida for their annual spring "rejuvenation." Planted on the beach each day, Kirby and family enjoyed the sand, water and afternoon "smoothies" with friends. Leaving picture-perfect weather, the Wilsons returned home ready to celebrate a very special day on April 15th ... Kirby's 13th birthday!

Dining on her ever-favorite pizza, Coke and cake, Kirby had no smiles as she used the two-fisted method of eating. But, as always, after she had had her fill, she acknowledged her adoring family and friends who surrounded her with smiles. "It is a blessing for us to see Kirby continue to recognize and enjoy her favorite meals with that heartwarming smile that inspires us to continue with a very special mission," Sue comments. "Every day of her life is a celebration of her incredible tenacity, for which we give thanks."



Our little teen angel



Some happy sweethearts

different. We were so pleased to see the many familiar faces showing their steadfast commitment to Kirby and the entire staff of The Drake, who worked tirelessly to ensure the success of the evening. This group affirms our belief that Kirby remains a viable child and year after year, empowers our strength and determination to find a cure for Sanfilippo. Brad and I will be forever grateful for their compassion."

FUNDRAISING NEWS

A "Sweetheart" of a Night

February 6th marked the Ninth Annual Sweetheart Dinner Dance at The Drake Hotel in Chicago, which was attended by 233 "sweethearts" who helped raise more than \$75,000 for research. The Drake Hotel again honored the event with its own "Red Day," where employees wore red to signify their support of the Foundation and its mission.

Sue Wilson states, "I have always said that this fundraiser represents a wonderful beginning to each year for us, and this year was certainly no

FUNDRAISING NEWS



Allison selling for the Cure

The Foundation Gives Thanks...

To Kirby's cousin **Allison Pratt** and all the members of **New Trier High School's Social Service Club** for making the Foundation the benefactor of the club's annual fundraising sale. This year, the club sold pizza (Kirby's personal favorite) and carrying bags over a two-week period to raise funds to support a "Cure for Kirby." Our thanks to the club members for their great planning and sales effort and to the faculty and students who bought the goods!!

To the members of **Miami University's (Ohio) Chapter of Alpha Lambda Delta** for choosing the Foundation as the benefactor of Jerry Springer's address on freedom of speech at the University, as well as thanks for the personal contribution from Mr. Springer himself. Now you're talkin'!

To board member **Charlie Notarus** and co-worker **Ken Pavitt** of Great Lakes Graphics, Inc. for choosing a donation to the Foundation in lieu of Christmas gifts for their clients. A very special gift indeed!

To **Lyons Township High School's winter cheerleaders** for contributing the proceeds from their lion auction to the Foundation. Three cheers for a group of young ladies who really know how to roar!

To **Lyons Township High School's Interact Club members** for choosing the Foundation as one of the benefactors of the 9th Annual Leon Eich Memorial Volleyball Tournament. Club members ask students and faculty to form teams, create their own team names and uniforms and solicit pledges for the charity. Prizes are given to the teams with the best spirit, most funds raised, best costume, and of course, the

winning team. This year 24 teams competed in this event. We can dig it!

To **Sharon Gleich** for donating the proceeds from her PartyLite party to the Foundation. It sure did brighten our day!

To **White Hen Pantry, 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, your "hometown girl" thanks you from the bottom of her little heart.

To our dear friends at **Highlands Presbyterian Church** for their continued love and support of our family and Foundation.

To **Eleanor Rounce** for her most generous support of the Foundation in fond memory of her husband and the Wilson family's minister and dear friend, **Rev. Robert Rounce**.

To **Randy and Barbara Bolduc** for their contributions to the Foundation in memory of dear friends **Michael Houlahan, Dianne Frazee and Stephanie May**.

To **Lynn Regnery** for her donation in fond memory of **Eric Johansson**.

To **Joanne Wilson** for her donation in memory of dear friend **Marion English**.

FUNDRAISING NEWS

News From Connecticut



Jacqueline with a great smile for the camera

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

To **Greg and Toni Graham** of Salinas, CA, for their most generous support of the Foundation in honor of their daughter, **Jacqueline**, who will turn nine years old on June 28th. Happy Birthday to Jacq!

Our thanks to **Lette K. Nocea** for her donation in honor of her "beautiful cousin," **Tessa Beachem**.

To the **IPA Wives Club** for donating the proceeds of its fundraiser to the Foundation in honor of **Ashleigh and Brooke Kidwell** of Lexington, KY.

The Logans are truly grateful for all the continued support. The greatest believer of Rhianna's Hope appears to be Rhianna herself. Each morning she awakes with a determination to stay strong. She goes to school and works very hard. She continues with her therapeutic horseback riding, which always brings her tremendous smiles and great pride, and she is planning her fifth trip to see her dolphin friends this October. Rhianna sends smiles to many each day. Hope remains strong in Connecticut.

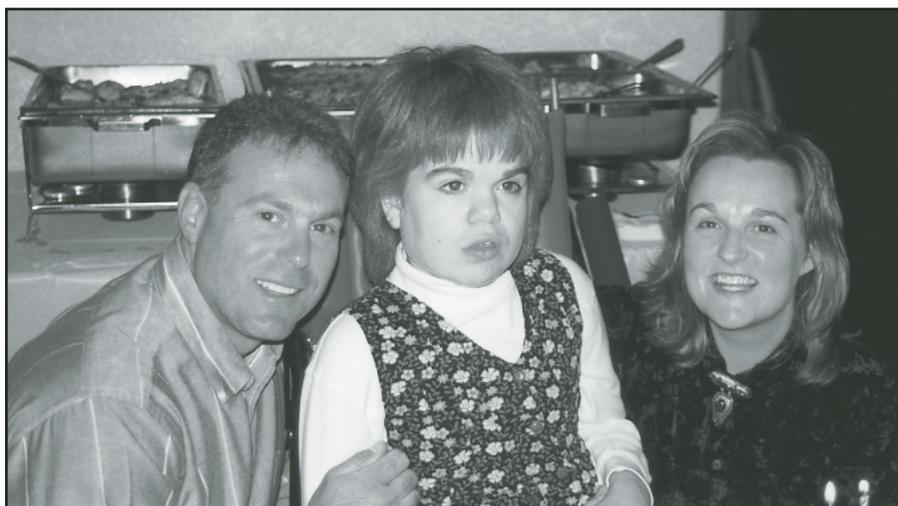
Our hats go off to **Louis Toffolon School** for the continued support of the Foundation with its "Hat Day" fundraiser held this past March in honor of Rhianna Logan.

The **Artisan's Marketplace** shone with its annual holiday fundraiser, "Stars for Hope." Many thanks for the hope you help to keep bright!

Justin and Tabitha Manafort celebrated the baptism of their first child by giving godparents, **Quinn Wazorko and Jason Manafort**, a special gift "as a thank you for playing such an important role in our daughter's life." The Foundation received a donation celebrating this very special occasion and these very special people.

Judith Morris of Tastefully Simply Gourmet Food donated a portion of her profits from an event held at Generations of Design, a longtime supporter of the Foundation. What a delicious idea!

And last, but certainly not least, our thanks to **Cory and Donna Gabel**, Rhianna's aunt and uncle, for their ongoing donations of a portion of the proceeds from Cory's solo piano instrumental CD, "One Road," to the Foundation. Sales continue, as do donations to the Foundation. Interested in this new "spin" to help? Just contact the Foundation to obtain a copy or log on to Cory's website at cdonavan.com.



Rhianna, surrounded by her proud parents, Gene and Cynthia, on her 11th birthday

RESEARCH UPDATE

Svitlana Garbuzova-Davis, Ph.D., D.Sc., Center for Aging & Brain Repair, Department of Neurosurgery, University of South Florida

In the last few years, our research group, led by Dr. Paul R. Sanberg, University of South Florida, has studied the potential of human umbilical cord blood stem cells to treat various neurodegenerative disorders. Data suggest that these cells are able to differentiate into pre-neural cells, making repair of an injured or defective nervous system possible.

In a previous study of human umbilical cord blood (hUCB) cell cultures, we found the Naglu enzyme (the enzyme missing in Sanfilippo B patients) was released by the cells, suggesting that hUCB cells could supply missing enzyme to reduce heparan sulfate accumulation (a consequence of Naglu deficiency). We followed these results with a long-term cell transplant study in a mouse model of Sanfilippo B for delivery of the deficient Naglu enzyme. After cells were transplanted into the lateral ventricle of the brains of 1-month-old mutant mice, a decrease in stereotypical disease-related activity and an improvement in cognitive function were noted in the mice, mostly in females. The long-term survival, migration, neural differentiation and morphological changes of hUCB cells, the normal appearance of neuronal architecture in the brain, and the reduction of heparan sulfate in the liver of treated mice all indicate the beneficial effect of hUCB cells. However, we supposed that administration of hUCB cells into the circulatory system of mutant mice could be more advantageous and might lead to new strategies of enzyme replacement for Sanfilippo.

The aim of our studies was to determine the effect of intravenous transplantation of hUCB cells into a

mouse model of Sanfilippo type B at different stages of disease. Fifty-four mice were administered hUCB cells at 6 months of age (**first study**: late stage disease), while sixty mice received hUCB cells at 3 months of age (**second study**: early disease symptoms). Non-treated mutants and normal mice were used as controls. At monthly intervals for 3 months (**first study**) or 6 months (**second study**) post-transplant, the animals participated in behavioral tests of cognitive function (active avoidance) and spontaneous movements (open field).

Mutant mice showed cognitive impairment in initial learning tests as early as 3 months of age versus normal mice. Transplanted mutant mice in each study showed cognitive improvement and a decrease in disease-related hyperactivity even when cells were administered in end-stage disease. These results demonstrate that intravenous administration of hUCB cells into Naglu mice at different stages (early symptomatic or end-stage) of disease has a beneficial effect, probably due to enzyme delivery into the enzyme-deficient mutant mice. In our initial study we were able to identify transplanted hUCB cells within many organs, including the brain, of mutant mice receiving hUCB cells. We also noted increased monocytes/macrophages in the peripheral blood; these particular cell types may assist in clearing the accumulated heparan sulfate from the body.

The data collected so far are very encouraging. However, in order to fully evaluate the effectiveness of hUCB treatment in Sanfilippo, it will be necessary to examine such important variables as gender differences in outcome and structural analysis of the affected organs in the proposed project.

**Robert K. Yu, Ph.D., Med.Sc.D
Institute of Molecular Medicine
and Genetics, Medical College
of Georgia**

Current research in Dr. Robert Yu's lab deals with the development of a cell transplantation therapy for Sanfilippo Syndrome. At present, there is still no effective treatment available to these patients. The approach we are undertaking involves the use of human embryonic stem cells that can be transplanted into brain to correct some of the metabolic defects. Stem cells are self-renewable and capable of becoming various cell types, such as brain, muscle, bone, and blood cells. More importantly they are capable of differentiating into several types of nerve cells, such as neuronal and glial cells.

Our goal is to transplant these cells into the brain of animal models with Sanfilippo to see if they can correct the storage of heparan sulfate. To achieve this goal, we first want to study the biological properties of these cells, first in test tubes, and then in brain. This basic knowledge is essential because we want to avoid abnormal growth of these cells, e.g., tumor growth. Since we hope to take advantage of the enzymes that are present in these cells, we will find out if these cells indeed secrete the missing enzymes that can produce the beneficial effect. We wish to test if these cells can make the correction, and if so, what type of cells is the best. Once we obtain the desired cells, we will transplant them into animals with Sanfilippo and examine the effects on the reduction of the stored materials and the long-term consequences. We also plan to engineer these cells to give them a boost of the specific enzymes to improve efficiency. Our long-term goal is to correct and alleviate the storage defects in Sanfilippo patients, using the cell therapeutic strategy.

FUNDRAISING OPPORTUNITIES

A Match For Kirby

Does your company have a matching gift program? It could double your support of the Foundation. Our thanks to **C.N. Nicholson and Altria Group, Inc., Kimberly Michaud and the Illinois Tool Works Foundation and Matthew Reid and the Ryan Foundation** for their perfect match!

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! Our United Way Chicago "Agency Number" is 3025558. For those of you out of state wishing to participate, please supply your local United Way agency with the Foundation name, address and our Federal ID #36-4033667.

Our thanks to **the employees of SBC, Wellpoint Health Networks, Inc. and United Parcel Service** for their pledges toward our mission.

Give Kirby Security

Tired of taxes? The Foundation now has a brokerage account available which 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 Brad or 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 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.

SAVE THE DATE

10th Annual Sweetheart Dinner Dance

Plans will soon be underway for the 10th Annual Sweetheart Dinner Dance to be held Friday, February 4, 2005, at The Drake Hotel in Chicago. Reservations will be \$200 per person and must be made in advance. Please contact the Foundation at (708) 784-0631 for further details and to learn how you can help to make it a "sweetheart" of a night for Kirby and others like her.

The Children's Medical Research Foundation, Inc.

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We're Short

Being short can be a good thing unless it means we can't fund all the research we'd like to this year. Not unexpectedly, donations to charitable causes are down this year, and those to the Foundation are no exception. Grant proposals are coming **due this month** for research projects that have shown great promise. Please help us to make up the gap between available grant money and projected research funding needs. Your generosity will make all the difference.

Please send donations now to:
The Children's Medical Research Foundation, Inc.
P. O. Box 70
Western Springs, IL 60558

Kirby and all the children afflicted with Sanfilippo thank you.



They're short, but cute!

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