



KirbyGram

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

June 2008

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

April 15th was a beautiful day for the Wilson family as Kirby turned 17 years old. Brad and Sue are thankful to be able to celebrate another year with her still able to enjoy favorite foods of pizza, Coke and cake.

As Kirby continues to endure the daily distress that her uncontrolled movements and spasms cause, Brad's and Sue's awe of her quiet resolve to enjoy the "in-between moments" continues to grow and inspire their determination to find comfort for their child. Sue explains, "Over the years, Brad and I have watched Sanfilippo slowly rob our child of abilities, and Kirby has always been the one to show us how to find happiness, focusing on the many blessings that remain in our lives. But watching her contend with the spasms that some days give her only minutes without discomfort is something we will never grow accustomed to watching. As she perseveres, we remain determined and hopeful to find a comfort for Kirby and a cure for Sanfilippo."



"Although this part of our journey tends to keep beating us down a bit, we still see many wonderful things around us," Sue continues. "There is Kirby's classroom staff at Lyons Township High School that continues to provide her with opportunities to maintain abilities, as well as search for ways they, too, can give comfort. There are the doctors and scientists we have working to find treatments and the cure. And last but certainly not least, it is spring, with all its warm sunny days bringing renewed life and bright, colorful flowers for us to walk among and enjoy with Kirby by our side."



The birthday girl.

FUNDRAISING NEWS

Marquette University's Student-Athletes Strike Gold Again for Kirby

For the second year, Marquette University's Student-Athlete Advisory Council (SAC) chose to fundraise for the Foundation in honor of fellow member Maggie Wilson's little sister, Kirby. The Council planned the "Beating the Buzzer for Sanfilippo" fundraiser for the January 29th Golden Eagles men's basketball game. A video clip of the Foundation created by the students was presented during the game at the Bradley Center, and over 40 student-athletes canvassed the crowd at the conclusion of the game to collect just under \$4,000 for the Foundation. Sue Wilson comments, "It is a wonderful feeling to realize the commitment of a group of young adults, who know Maggie and the plight of her sister, to help make a difference in the lives of children like Kirby. As I have said before, it is their compassion and energetic can-do attitude that not only amazes us but also makes us proud to have a daughter be a part of this giving environment."

Sweet Night at The Ritz

February 8th marked the Thirteenth Annual Sweetheart Dinner Dance at The Ritz-Carlton Chicago. It was the Foundation's second year with The Ritz, and all 175 guests were treated to a champagne cocktail reception followed by a four-course dinner from its award-winning kitchen. After dinner, guests opened up their hearts for

Kirby with their enthusiastic participation and record-setting bidding in a live auction. Of course, the evening would never be complete without Michael Lerich and his group of talented musicians who have entertained the Foundation's sweethearts each and every year. This year's event raised just under \$62,000. A very sweet night!



Brad Wilson (r) with niece Molly and friends.



Brad and Sue with Maggie, her tennis team, coaches and fellow student athletes.

FUNDRAISING NEWS

A Note From a Foundation Friend

When my wife, Karen, and I were kids, our parents and grandparents would occasionally give us various stock certificates, which were placed into a brokerage account. A very generous but random form of giving. Over the years we had accumulated a nice sum of money with these gifts. That's the good news. The bad news is that we really had a problem when we wanted to sell the securities since we had no idea of the cost basis of the stock. Cost basis is the value of the stock on the day you were given the security. As you may know, in order to figure the capital gains tax on a stock sale, you need to know the sale price and cost basis of a stock. A friend of mine advised me to just guess at the cost and see what the IRS would say. I took another route.

Here's a perfect solution. Gift your stocks without a known cost basis to a charity such as The Children's Medical Research Foundation. You get to deduct the full value of the stock as posted on the day of the transfer. There's no need to find or report a cost basis. Problem solved and we feel great knowing that one of our favorite charities is putting the money to good use. The process is painless since your broker will do all the work. Sue Wilson can provide the account information necessary to transfer any stocks. It's painless.

I'm not a tax accountant so check with one.

Bill Rajki

The Foundation Gives Thanks

To **Molly Russell**, Kirby's cousin, for her November 6th fundraiser at Max & Erma's in Burr Ridge. She really knows how to raise money and spread the word about Kirby and the Foundation!

To **Bill and Karen Rajki** for their most generous donation of stock and "**Giving Kirby Security.**" What a great gain for the Foundation!

To the employees of **Crowe Chizek and Company** in Oak Brook for going "**Kasual for Kirby,**" and raising \$550 for the Foundation. Blue jeans never looked so good!

To board member **Charlie Notarus** of Great Lakes Graphics, Inc. for also choosing a donation to the Foundation in lieu of holiday gifts for his clients. A very special gift indeed!

To **Margaret Dawe, Nicholas Megofna and the anonymous donor** from Adams & Knight who designated the Foundation as their charity of choice in their employer's United Way campaigns. Thanks for uniting for Kirby!

To the many contributors who used the donation envelopes as an opportunity to give to the Foundation. Donations from our December newsletter totaled \$4,220!

To **Ralph H. Kaiser** for his donation in honor of **Sydney and Hunter Moff** of Williamsport, PA.

To **Rob and Alison Credit** for their donations in celebration of

the births of **Shea Ward, Delila Henley and Madelyn Richner.**

To **Bob and Marcia Kaplan and Mr. & Mrs. Howard Feuer** for their donations in celebration of the birth of Juliette Anne Zabel.

To **Lynn Regnery** for her donation in honor of her sister, **Gretchen Wallrich**, and in memory of their mother, **Verla Regnery.**

To **Barbara Bolduc** for her donation in memory of longtime friend, Jim Address.

To **Fred and Beth Angeli** for their donations in memory of **Roger Untalan and Janice Kolbas.**

And to **Rosario Rizzo** for his donation in fond memory of his wife, **Doris A. Rizzo.**

To The Families Working Together For The Cure

News From Connecticut

The Artisan's Marketplace continued its holiday tradition with the "Santa for Hope" raffle and "Stars for Hope" fundraisers. This year, thanks to artist



Rhianna enjoying music with friends Marissa and Michael.

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Maria-Lucia Sileo, who donated a portion of her hand-painted ornaments profits, the shop was able to add “Ornaments for Hope” to its fundraising efforts and raised \$1,300.

Emily Anderson, Rhianna’s friend from pre-school, and her grandmother, **Nancy Anderson**, made cash jars with Rhianna’s picture and story on them for Emily’s parents’ work places. Donations totaled over \$260. Friendship is a wonderful gift!

A donation was received from the **Connecticut Area Classic Thunderbird Club** in honor of Rhianna. The Foundation thanks this group for its steadfast support of “Rhianna’s Hope” and our mission.

Donations were also received in Rhianna’s name from **Peter and Joanne Brandien** and **Picture This Custom Framing**.

A Tisket, A Tasket.....Look, It’s Anna Creating Baskets!

Anna Kidwell, mother to **Brooke and Ashleigh** once again made the drive from Kentucky to



Brooke graduates and moves on to join Ashleigh in high school.

Chicago this past February to attend the Sweetheart Dinner Dance. As usual, her car was filled to the brim with items she collected throughout the year for the auction. The morning of the event, you typically can find her busy on the floor, sorting through the items to create beautiful baskets for the auction. This year, the only difference was she managed to slip in an amazing handcrafted, wood birdhouse as a surprise for Brad, direct from the creative hands of her husband, **Dave**.



Ashleigh

Not stopping there, Anna’s fellow member **Bev Sweeny** of **IPAWC (Independent Pilots Association Wives Club)**, turned a holiday gathering for members into a fundraiser for the Foundation, raising over \$6,900. The Foundation gives thanks to all the club’s members for sharing their holiday cheer!

Grace For A Cure Makes Strides

John and Joan Bellontine of Huntington Station, NY, formed **Grace for a Cure** in honor of their daughter, Grace. Their first 5K walk raised over \$10,000,



Grace setting the pace.

which they shared with the Foundation as well as two other fundraising organizations that focus on research and awareness, Ben’s Dream and the National MPS Society. We thank John and Joan for including the Foundation as one of the benefactors of their walk for the cure.

Speaking of Working Together

The Children’s Medical Research Foundation is proud to announce its participation in a Partnership Grant with Ben’s Dream and the National MPS Society. One \$60,000 grant will be issued to support basic or clinical research for Sanfilippo. The grant is for a two-year period, with half of the total funding beginning July 1, 2008. The Society and its advisory board have received all applications and will be responsible for awarding the funds. It is an honor to be working with both the Society and Ben’s Dream to promote this most important research.

RESEARCH UPDATE

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

Development of Stem Cell Transplantation Therapy for Sanfilippo Disease: Where Are We Now and Where Are We Going?

Dr. Yu and his team have continued their effort in developing a cell transplantation therapy to alleviate the devastating effects of Sanfilippo disease (Mucopolysaccharidosis III, MPSIII). Toward this end, they have employed neural stem cells (NSCs) for transplantation. There have been strong efforts worldwide in promoting these cells for the treatment of a variety of neurodegenerative diseases because stem cells are capable of becoming neural cells and integrate into the damaged brains once transplanted. One major uncertainty facing investigators in the field, however, is to ensure that these cells behave normally and do not turn into malignant cells. During the past year, Dr. Yu and his team have learned much about the properties of the NSCs in test tubes. This basic knowledge is essential because Dr. Yu needs to harvest enough corrective cells for transplantation. Once the cells are transplanted, he wants to make sure the donor cells survive, migrate, and grow normally within the recipient's brain, and to avoid abnormal growth of these cells, e.g., tumor growth. Dr. Yu and his collaborators have already transplanted these cells into the brain of animal models with MPSIII to see if they can correct

the storage of heparan sulfate. Preliminary studies have shown that these cells grow after transplantation. More importantly the recipient animals survive the transplantation surgery for a prolonged time. Animals with MPS appear to have improved biological function, as assessed by behavioral tests, which is very encouraging.

Although there have been attempts at using gene therapy for MPSIII, there have been mixed results, especially when virus-based techniques are required. Several reports have appeared on the appearance of serious side effects. During the past year, Dr. Yu and his team have started to explore a new technology for gene therapy, the so-called "Sleeping Beauty" or "transposon" technology. This is a novel way to replace missing or defective genes into host chromosomes. This technique is gaining momentum in the gene therapy field because of few side effects and the ability to integrate the missing genes into the host chromosomes. Embryonic stem (ES) cells may also be amenable to ex vivo engineering by DNA modifications that render them more suitable for gene therapy. Preliminary results from Dr. Yu's laboratory have shown that it is feasible to fortify ES cells with enzymes that are missing in MPSIII using this technique. He plans to transplant these engineered cells into animal brain in an attempt to restore function in the diseased tissue. He is keenly aware that the safety and efficacy of these strategies must be fully evaluated before their use in humans with MPSIII.

In preparation for use in humans, Dr. Yu has established the first state-supported Umbilical Cord Blood Stem Cell Bank in the State of Georgia. Cord blood is rich in stem cells that can be harvested for clinical use. This bank is supported by funds provided by the State of Georgia and the Medical College of Georgia. In fact, the State of Georgia has reaffirmed its commitment to cord blood stem cell collection and research by appropriating additional funds to support this facility for 2009. In 2007, Governor Sonny Perdue signed a bill establishing the Commission of Cord Blood Stem Cell Research and Medical Treatment and appointed Dr. Yu as a member of the Commission. The Commission delivered its recommendation in December. To continue this effort, Governor Perdue has reappointed Dr. Yu as one of the members of the Commission of Saving the Cure in April, 2008. The purpose of this Commission is to provide guidance on state support for clinical and basic research in stem cells in Georgia. We anticipate that we will have sufficient stem cells for the treatment of a variety of diseases, including Sanfilippo disease, in the near future.

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

During the last few years, our research group at the University of South Florida has studied the potential of human umbilical cord blood (hUCB) stem cells to treat

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RESEARCH UPDATE

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various neurodegenerative disorders (stroke, ALS, Parkinson's disease) as well as brain and spinal cord injuries. Data suggest that these cells are able to repair a damaged or defective nervous system.

In Sanfilippo syndrome type B, a deficiency of the *Naglu* enzyme leads to accumulation of heparan sulfate, a major feature of this disease. Our previous results demonstrated that a single administration of hUCB cells into the veins of Sanfilippo mice at early or late stage disease had a beneficial effect, probably due to enzyme delivery into these enzyme-deficient mutant mice. The observed behavioral improvements, indicating advantages of the administered hUCB cells, were supported by the survival, distribution, and development into different cell types of the transplanted cells. After administration of hUCB cells, the cells were found widely distributed within and outside the central nervous system. Also, heparan sulfate accumulation was reduced in the liver and spleen of *Naglu* mice, mainly in females, 6 months after receiving hUCB cells. Additionally, an anti-inflammatory effect by hUCB cell transplantation was determined.

However, most observed behavioral benefits in Sanfilippo mice were limited to the first months after transplantation, possibly due to declining production of the missing enzyme. To address this possibility, we investigated the effect of repeated hUCB cell infusions. We showed that multiple administrations of hUCB cells into mutant mice improved behavioral activity of

both sexes for a prolonged period. The behavioral results were supported by detection of transfused hUCB cells in the blood circulation and peripheral organs of mutant mice; increasing cell numbers were noted after each cell injection. Another important advantage of repeated hUCB cell injections was significantly reduced heparan sulfate accumulation in the liver of *Naglu* mice compared to singly treated or non-treated mutants. Thus, our results demonstrated that multiple administrations of hUCB cells into *Naglu* mice at an early symptomatic stage of disease have a beneficial effect compared to a single injection, probably due to continuous enzyme delivery into the enzyme-deficient mutant mice. However, the potential for hUCB cell migration to the brain and identification of cell types developing from the cells grafted into mutant mice still need investigation. The objective of the proposal was to determine whether hUCB cells injected into a vein migrate to the brain and develop into appropriate cell types.

The current study demonstrated that previously described benefits of multiple hUCB cell injections into *Naglu* mice probably occur from migration of the administered cells to the various structures of the brain, as indicated by the larger number of migrated cells found in repeatedly vs. singly treated mutant animals. The importance of this finding is probably continuous enzyme delivery to the brain due to the higher numbers of surviving cells after multiple injections;

consequently, multiply treated animals show more neurological improvements than animals receiving a single dose of hUCB cells. The hUCB cells were found widely distributed among different brain structures, mainly in the hippocampus, a brain structure responsible for short term memory, formation of the new memories and spatial navigation. However, the multiply treated mice produced fewer cells able to develop into neurons or astrocytes compared to mice receiving a single injection of cells. Based on these results, it is possible to conclude that **continuous enzyme delivery** into enzyme deficient *Naglu* mice by repeated MNC hUCB cell administrations, rather than cell development to another cell type, might be more promising in developing a cell-based therapy for Sanfilippo Syndrome type B.

The data collected so far are very hopeful; however, our current project addresses a number of areas needing further study. We will investigate the migration route of administered cells to the brains of mutant mice. First, we will determine the condition of the blood-brain barrier in Sanfilippo. Determining when, where, and if, this barrier is damaged in Sanfilippo is important not only for examining transplanted cells' migration, but will also be crucial to increased understanding of this disease and developing a future drug-based treatment.

FUNDRAISING OPPORTUNITIES

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.

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.

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.

In Fond Memory

Joe Langford of Johns Island, SC, 13-year-old son of Frank and Sissi Langford and brother to Jessie and Maggie, passed away peacefully on April 26th surrounded by his family and friends. He was diagnosed with Sanfilippo in 1999 along with his sister Maggie.

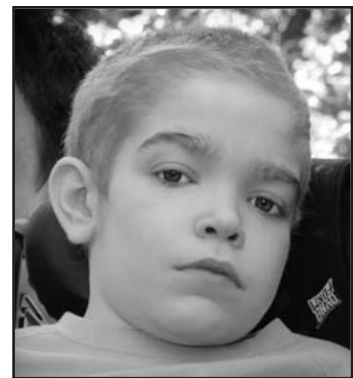
His family describes his love of life that he shared with all so fortunate to know him. "Joe Langford touched the hearts and lives of many teachers, therapists, caregivers, doctors and nurses in Charleston. Joe taught so many people the definition of courage and the importance of looking past their own problems to make the best of their situation. Joe was a quiet, thoughtful and loving little boy who always made people smile. He loved being around all the people in his life. Joe lived a very exciting, meaningful and successful life."

His accomplishments were many . . .

In addition to attending school for seven years, he participated in therapeutic horseback riding, swam,

attended day camp, and through the Make-A-Wish Foundation, traveled to Disney World in Florida. He was at Centennial Olympic Park in Atlanta when a brick was placed in the park in his honor prior to the 1996 Olympic Games. He raised funds for research through his participation in MPS walks and dramatically increased awareness through his meetings with U.S. Senators, Congressmen and Congressional staff members and was featured in Congressional testimony as well as newspaper articles. In December, 2007, he attended his sister Jessie's graduation from Georgia Tech in Atlanta.

Our deepest sympathy to Frank, Sissi, Jessie and Maggie for their loss.



Joseph Francis Langford.

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Western Springs, IL 60558

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