



# KirbyGram

June 2007

*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

Kirby turned 16 years old this past April and as always, her loving family and close friends surrounded her. Sue states, "We are extremely grateful to have Kirby with us, enjoying her favorite foods of pizza, Coke and cake, but as I have said in the past, I am reminded of the words a father wrote on his days in a hospital observing families of critically ill children. He stated it was a powerful experience, and he sensed 'a collision of the parents' hopes and fears.' Now, more than ever, these words give incredible insight into Brad's and my emotions, as well."



**Sweet 16 and very cool!**

Kirby continues to experience daily the anxiety and discomfort from her uncontrolled movements and spasms. Brad and I can only watch as she fights to regain control. Our fear is now very real and tears at our hearts. But as the past 16 years have taught us, it is Kirby who shows us the way of hope. She has risen to new heights of strength and courage that we thought not possible, rarely vocalizing her discomfort. She perseveres and can now sit on her own for short periods and responds to our words and touch. It is those denim-blue eyes looking into ours and the feeling of her hand holding on tight that comforts our hearts. She is a powerful force that defines characteristics we can all aspire to. Brad and I are honored to call ourselves her parents and will never give up our search for comfort and a cure for Kirby and others like her.

## FUNDRAISING NEWS

### Puttin' On The Ritz

There were 174 sweethearts who attended the 12th Annual Sweetheart Dinner Dance on February 9th at The Ritz-Carlton Chicago. Thankfully, this year's event turned out to be a perfect evening for all. Guests were treated to a champagne cocktail reception followed by a signature four-course dinner from The Ritz's award-winning kitchen. After dinner, guests opened up their hearts for Kirby with their enthusiastic participation in the Foundation's first ever 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 guests for the past twelve years. The dinner dance raised over \$73,000 for the Foundation, thanks to the generosity of the event's sponsors and attendees.

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

Marquette University's Student-Athlete Advisory Council learned of a freshman tennis player whose little sister's affliction inspired a Foundation that was formed to raise funds for a cure. The Council decided it, too, could help. Yes, that tennis player would be Maggie Wilson, Kirby's big sister. The Council planned the "Beating the Buzzer for Sanfilippo" fundraiser for the February 19th 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 on getting the news of the Council's decision, "It is an incredible feeling to realize a group of young adults, just getting to know Maggie, learn of her sister, and their first thought is 'we can do something about this.' It is their compassion and energetic can-do attitude that not only amazes us but also makes us ever so proud and privileged to have a daughter be a part of this giving environment."

### The Foundation Gives Thanks....

To the "C115 Staff," the Lyons Township High School classroom staff working with Kirby, for their kind letter to her classmates' parents last December asking they consider a gift to the Foundation as a alternative to staff gifts. We greatly appreciate their thoughtfulness and the many donations received in celebration of the holiday and their very special work with some very special children. Not stopping there, the school's

**Functional Program Staff** made personal donations as well!

To **Gresham, Smith and Partners**, of Nashville, TN, for their most generous support of the Foundation this past holiday season as they chose "to celebrate the season of giving" with a donation to the Foundation in honor of their business friends and associates.

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 **Mr. & Mrs. Joseph Sanfilippo** for their Christmas donation in honor of **Mr. & Mrs. Orval Adam**.

To **Anthony Cefali**, his fellow Locke band members and all the other bands who participated in the December 22nd "Christmas Extravaganza," to raise funds for the Foundation. Over \$632 was raised....rock on!!

To the employees of **Crowe Chizek and Company**, in



**The Wilsons accept a check from representatives of the Student-Athlete Advisory Council, as well as the women's tennis team.**



## FUNDRAISING NEWS

Oak Brook, for going “Kasual for Kirby,” and raising \$550 for the Foundation.

To **The LaGrange Highlands Woman’s Club**, which over the years has repeatedly chosen the Foundation as one of its benefactors. Contributions now total over \$15,300! “These women all live within our community, and I am honored to have them as friends and neighbors. As mothers themselves, they have never forgotten dear Kirby or the importance of our mission. Brad and I are grateful for their prayers and support,” says Sue.

► To **Hunter Douglas Hospitality** in Chicago and the **Wellpoint Foundation** in Princeton, NJ, for their matching gift donations. A perfect match for Kirby!

► To **Margaret Dawe, Colleen McClaran, Nicholas Megofna**, and the anonymous donors who designated the Foundation as their charity of choice in their various employers’ United Way campaigns. We appreciate them uniting for Kirby!

► To the employees of **Bank Financial** in Burr Ridge for choosing the Foundation as the benefactor of “Charity Day,” during which employees make a donation to a designated charity.

► 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, your “hometown girl” thanks you from the bottom of her little heart.

► To **Don and Jan Matheny** for their gift to the Foundation in honor of **Dr. Marcy Lake**.

► To **Fred and Beth Angeli** for their donations to the Foundation in memory of **Jimmy Shovald and John Viitala**.

► To **John and Carol Rante** for their donations in memory of **Mr. J. Lenc and Mr. J. Bisset**.

► To **Joanne Wilson** for her donation in memory of **Kent White**.

► To **Drs. John and Margaret Jones** for their donation in memory of “**Pinky**” **Wilburn**.

► And, to the many friends and family of **Walter Rajki** for their donations in his memory.

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

Syosset High School in Long Island, NY, organized a silent auction of students’ original artwork donated by the school’s AP design classes to raise funds for the Foundation in honor of Grace Bellontine of South Huntington, NY. Syosset’s Student Government sponsored the project and paid to mat and frame each of the 26 pieces. A local gallery donated the space for the event, which was run entirely by the students, while

local businesses donated food, drinks and raffle prizes. Other clubs within the school added their own donations, as well. A \$3,000 check was enclosed in a letter which described the students’ experience as “both powerful and inspirational” and expressed their “hopes and wishes for a cure soon.” We could not say it better ourselves!!

A donation was received from **Stephen and Besty Fowler** of Shelby Twp., MI, in honor of their daughter Kim, who celebrated her 19th birthday on December 30th.



**Kim Fowler and brother Jeff just after a 5K.**



**Joan and Grace Bellontine (second from left) along with Syosset High School staff and students.**

## News From Connecticut

**Cory and Donna Gabel**, Rhianna's uncle and aunt, continue their fundraising efforts on behalf of Rhianna for the Foundation. Their latest event last November celebrated the release of Cory's new solo piano recording, "Alabaster Sky," and raised \$1,250 for research! The Foundation continues to receive a portion of proceeds from his CD sales, which also include his first release, "One

Road." To learn more about Cory, his work and how to obtain CDs, contact the Foundation or log on to [cdonavan.com](http://cdonavan.com).

**The Artisan's Marketplace** holiday fundraiser continues to grow in popularity. The "Santa for Hope" raffle and "Stars for Hope" made \$1,755 for the Foundation. "Stars for Hope" is a craft kit for children put together right at the shop by Rhianna's family and her friends. Customers purchase the kit, allowing their

children to create the star and keep it, reminding them of Rhianna. Or they can return the star to the Artisan's Marketplace, where others can purchase it, doubling the donation to the Foundation. What a shining example of compassion.

**Debra Dawson** and her sister **Lee Longo**, of the **Hair Spa and Kids Cut** recently were offered a booth at a local children's conference and immediately thought of their own hometown girl, Rhianna, and the Foundation. The two did their own mini "**Cut Against Time**" and spent the day offering kids' haircuts at the conference, raising \$400 for the Foundation!

**Alan and Donna Theriault** are busy planning for the "**Rockin' for Rhianna**" annual dance and silent auction. This year, the event will take place in the fall at a new location. Stay tuned for details!



Rhianna enjoying a hug from her dad.

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## RESEARCH UPDATE

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

### Development of a Stem Cell Therapy for Sanfilippo Disease: Where We Are and Where We Are Going

Dr. Robert Yu and his team at the  
Medical College of Georgia

(MCG) have engaged in developing a cell-transplantation therapy to alleviate the devastating effects of Sanfilippo disease (mucopolysaccharidosis III). The stem cells are derived from the inner cell mass in the blastocytes at early embryonic stages of development. Recent studies indicate stem cells can also be isolated from other sources, such as umbilical cord

blood, amniotic fluid, and even adipose tissue. These primitive stem cells are self-renewable and multipotential, meaning they can develop into different cell types, such as nerve, muscle, and bone. For this reason, stem cells can be harvested and transplanted into damaged brain, where they then can repopulate damaged tissues. Transplantation therapy, therefore, represents a viable

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therapeutic approach for repairing injured brain tissue caused by many different neurogenerative diseases, including Sanfilippo disease, multiple sclerosis, strokes, Parkinson's disease, and so on.

For practical purposes, Dr. Yu has induced stem cells to differentiate into neural stem cells (NSCs) that are committed to developing into brain cells. Once these cells are transplanted into damaged brain, they can correct some of the metabolic defects of Sanfilippo disease (MPSIII). During the past year, Dr. Yu and his team have learned much about the properties of NSCs and how they populate and interact properly with each other in test tubes. This basic knowledge is essential because Dr. Yu wishes to make sure that he can produce enough cells for transplantation. Once the cells are transplanted, he wants to make sure that they survive, migrate, and grow normally within the recipient's brain, and also that they avoid abnormal growth (e.g., tumor growth). Dr. Yu and his team have already transplanted these cells into the brain of animal models with MPSIII to see if these cells can correct the storage of heparan sulfate. Preliminary studies have shown that transplanted NSCs grow after transplantation. More important, the recipient animals survive the surgery for a prolonged time. Animals with MPS appear to have

improved biological function, as assessed by behavioral tests, which is very encouraging.

Embryonic stem cells (ESCs) may also be amenable to ex vivo engineering by DNA modifications that render them more suitable for gene therapy. Using viral infection, Dr. Yu has successfully fortified ESCs with enzymes that are missing in MPS cells. He plans to transplant these engineered cells into animal brain with MPS 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 MPS.

In preparation for use in humans, Dr. Yu has established the first state-supported umbilical cord blood bank in the state of Georgia. Umbilical cord blood is rich in stem cells that can be harvested for clinical use. The bank is supported by funds provided by Georgia and by MCG, among other sources. In fact, the governor of Georgia, Sonny Purdue, has signed a bill establishing the Commission of Cord Blood Stem Cell Research and Medical Treatment and has appointed Dr. Yu as a member of the commission. Dr. Yu and his team anticipate having sufficient numbers of stem cells in the near future to investigate the treatment of a variety of neurodegenerative conditions, including Sanfilippo disease.

**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 various neurodegenerative disorders, 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. Our results demonstrated that a **single** administration of hUCB cells into the veins of Sanfilippo mice at different stages of disease (early symptomatic or late stage) had a beneficial effect, probably due to enzyme delivery into these enzyme-deficient mutant mice. These previously shown behavioral improvements, indicating advantages of the administered hUCB cells, were supported by our results on 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. Although many cells were associated with blood vessels, confirming that transplanted cells were still

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present in the blood circulation, some cells were found in the brain and peripheral organs. Our results also showed that 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 a declining production of the missing enzyme over time. To address this limitation, it was necessary for us to investigate the effect of **repeated** hUCB cell infusions during the disease over time.

The aim of our pre-clinical translational study was to determine the effect of **multiple** intravenous transfusions of hUCB cells into a mouse model of Sanfilippo type B. We investigated the ability of repeated hUCB cell administration to ameliorate/prevent behavioral dysfunction in mutant mice.

As we showed previously, the Sanfilippo mice demonstrated early hypoactivity (3 months of age, early symptomatic stage of disease) with impaired learning (mostly, in males) followed by

hyperactivity and memory impairment with aging (8-9 months of age) of both sexes. Multiple administrations of hUCB cells into mutant mice showed improved behavioral activity of both sexes with increased repeat cell transplants. Interestingly, learning improved in mutant *males* treated multiple times mainly at the 3rd and 4th months after cell grafting; whereas their ability to remember (memory) tended to improve at 4th and 6th post-transplant months. During learning, Sanfilippo *female* mice treated multiple times showed learning improvement at months 4 through 6 after cell injection, although their improved memory was observed early, in the 3rd month post-transplant. The behavioral results were supported by analysis for the presence of transfused hUCB cells in the blood circulation and peripheral organs of mutant mice, showing increasing cell numbers after each cell injection.

Additionally, neuronal architecture in the hippocampus, analyzed by using a sensitive Golgi staining of dendritic domain of the neurons, was improved in *Naglu* mice of both sexes after multiple administrations of hUCB cells. Another important advantage of therapeutic tactic of repeated hUCB cell injections was

that heparan sulfate accumulation was significantly reduced in the liver of *Naglu* mice after receiving multiple deliveries of hUCB cells over 6 months compared to single-cell-treated *Naglu* mice and non-treated mutants.

The data collected so far are very hopeful; however, our current project addresses a number of areas needing further study. We will investigate potential migration of intravenously administered cells to the various structures of the brain of mutant mice. Also, analysis of specific immunophenotypes of these transfused cells will be performed, since this is an important criterion for evaluating/confirming the nature of the significant beneficial effects of hUCB cells in Sanfilippo mice.

The **repeated** administration of hUCB cells into the blood circulation of Sanfilippo mice may lead to the development of a new strategy for enzyme replacement for Sanfilippo. Our therapeutic tactic of continuous delivery of the missing *Naglu* enzyme by multiple cell administrations may be critical to developing a cell transplant strategy.

## 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.



Chillin' with sister Maggie

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