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MSUD NEWSLETTER

Published by the MSUD Family Support Group • Volume 27, Number 1 • Spring 2010

MSUD Symposium 2010
June 24 - 26
Lincolnshire, Illinois

Hope to see you there!
See Page 3 for details...

DEVELOPMENT OF NEW TREATMENT FOR SOME FORMS OF MAPLE SYRUP URINE DISEASE

Brendan Lee, M.D., Ph.D., Professor, Department of Molecular and Human Genetics, Baylor College of Medicine

Over the past few years, we have tried to translate our experience in treating urea cycle disorders to developing new applications for maple syrup urine disease.

Sodium phenylbutyrate is a medication that has been routinely used to treat urea cycle disorders. We have observed that this medication can lower branched chain amino acids. While this side effect is important for its use in urea cycle disorders, we wanted to test whether it may be useful in lowering branched chain amino acids in MSUD.

In our preliminary studies in patients with MSUD, skin cells from patients with MSUD, and mice, we have discovered that phenylbutyrate may indeed be useful in lowering branched chain amino acids in at least some cases of MSUD. However, it is still too early to recommend its wide spread use.

(*New Treatment* cont. on page 2)

Medical Foods Equity Act

Sandy Bulcher, RN
Director, MSUD Family Support Group

There are approximately 20,000 Americans with Inborn Errors of Metabolism, and most require medical formula and low protein foods to treat their disorder. Currently, 38 states have laws governing medical formula and low protein food coverage. However, the laws vary significantly from state to state and many families are not able to fully benefit from the legislation due to restrictions, such as age. In addition, insurance plans governed by the Employment Retirement Income Security act, ERISA, are not required to follow state mandates, leaving many families unable to benefit from legislation passed in their state. This includes those covered by companies that have a large number of employees and are often self insured.

I've worked hard to get passage of legislation in Ohio that

(*Medical Foods* cont. on page 2)

Inside This Issue:

(*Medical Foods* cont. from page 1)

would require insurance companies to pay for metabolic formula, with no success thus far. I've also watched as parents from other states worked countless hours to get passage of legislation, only to be disappointed by the limitations added to the legislation that restricted coverage for many. To meet the needs of most, the answer seems to lie in federal legislation rather than state, which would require insurance plans to cover medically necessary formula and low protein food.

Finally the time has come, and federal legislation was introduced this past fall. Senator John Kerry of Massachusetts introduced the Medical Foods Equity Act, also known as S.2766, following recommendations of the Advisory Committee on Heritable Disorders in Newborns and Children. This legislation is also endorsed by the American Academy of Pediatrics, The Society of Inherited Metabolic Disorders, and the Genetic Metabolic Dieticians International. Many parent groups, professionals, and organizations support the legislation, including the MSUD Family Support Group.

Specifically, the legislation requires federal health plans, such as Medicaid, Medicare, CHIP, and Tricare, as well as, private health insurance to cover the cost of medical food (formula) and foods modified to be low in protein. The federal legislation would not be subject to ERISA guidelines, thus all insurance companies would be required to comply. According to the bill, the Secretary of Health and Human Services has the authority to determine age–specific minimum yearly coverage for all health insurance plans. The minimum yearly coverage applies to individuals across their entire life span. The secretary also has the authority to update minimum yearly coverage levels, according to the cost of living index and actual cost of treatment.

Senator Kerry is the sponsor of the legislation, but he needs our help to find co sponsors. A co-sponsor is a legislator who steps forward and demonstrates that he or she supports a bill by signing on as a co sponsor. The more co sponsors a bill has the more likely it is to pass. This is where we need your help. Please contact your state senators and ask them to sign on as co-sponsors of the Medical Foods Equity Act, also known as S. 2766.

Senators can be reached by phone or email. Your phone call or email will be addressed by one of the senator's aides, whose job it is to inform their senator of issues important to his or her constituents. To learn more about the legislation, go to the National PKU Alliance website at www.npkua.org, click on the "Legislation" tab, and "Talking Points".

Don't forget to ask your family and friends to contact their state senators too! Thanks for your help! ■

16th Annual Metabolic Camp

Join us June 21-26, 2010 for Metabolic Camp at Emory University in Atlanta, GA!

This is a model, research-based camp for young women 12 years age and older with PKU and MSUD, which focuses on building social support through a variety of activities including nutrition education, cooking classes, discussion groups, and local field trips. The camp typically accepts 30 attendees on a first-come, first-served basis. Registered dietitians from across the nation and around the world volunteer their time to serve as camp counselors, and nutrition students provide support as assistant counselors.

The cost of the camp is $325 per person, which includes all sponsored meals, lodging, group activities, field trips, and closing banquet. Partial scholarships are available for certain financial circumstances. Check with your local RD and clinic to see if local sponsorship is available.

For more information about the Metabolic Camp, visit www.metcamp.org and contact Rosalynn Borlaza (Camp Coordinator) at (404) 778-8521 or rborlaz@emory.edu .

(*New Treatment* cont. from page 1)

We are planning to perform a definitive clinical trial to test the effectiveness of phenylbutyrate in MSUD, and to determine whether we can develop tools to screen for patients who might respond to its use. We don't expect that this treatment can cure MSUD. Instead, we hope that it may be a useful treatment in combination with standard dietary therapy. Together we hope that this may decrease frequency and severity of illnesses as well as enable relaxation of dietary restriction.

If you would like to participate in a web conference to learn more about Dr. Lee's study, contact Sandy Bulcher at 614.389.2739 or dbulcher@aol.com. This will be held on Wednesday, Feb. 24th at 7 pm CST. Participation is limited to 50 people so interested individuals should respond quickly. Dr. Lee will also be presenting at the MSUD Disease Family Support Group Symposium on June 24-26. ■

MSUD Symposium June 24 - 26 Lincolnshire, Illinois

With the bitter cold all around us, it's time to start thinking about your summer vacation. This year come and explore Chicago and attend the 15th Bi-annual MSUD Symposium at the Lincolnshire Marriott Resort.

The resort is only 25 miles from O'Hare International Airport and 18 miles north of downtown Chicago. The resort has indoor and outdoor heated pools with bike trails, golfing and many other activities right on the property. Our meals will be served at the hotel's picnic grove, which has a playground and other activities for the kids.

An exciting agenda is planned. Speakers include Dr. Lee from Baylor College of Medicine, who will speak on his research exploring the use of Buphenyl to treat MSUD. There will be a twist to our regular nutrition segment with Chef Jeff, who will be making recipes that can be shared by the whole family. We will also be discussing ways in which you can help the MSUD community by learning some easy fundraising ideas. A number of other topics and speakers will be covered.

The room rate is only $104. The hotel will honor this rate for 3 nights before and after our event. Simply copy the link below and include it with your electronic correspondence to facilitate the reservation process. You will be directed to the property's home page with the code already entered in the appropriate field.
All you need to do is enter your arrival date to begin the reservation process.

http://www.marriott.com/hotels/travel/chiln?groupCode=msumsua&app=resvlink&fromDate=6/24/10&toDate=6/29/10

So start planning your summer now. Join us on Thursday, June 24th to meet new and old friends while sharing information, networking, and socializing. Watch for the brochure which will arrive in your mailbox in the spring. All the information will be updated on the MSUD website **http://www.msud-support.org/**

For more information, contact: Barbara Mudrick at barbmud@yahoo.com or 224-944-1282

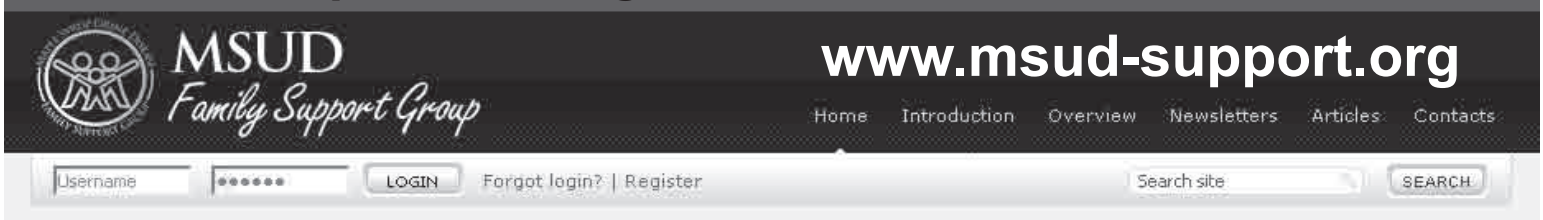
Hope to see you there!

Travel Assistance to attend Symposium 2010

The United Services Foundation again graciously supplied funds for the MSUD Family Support Group to help families and individuals with MSUD who need financial assistance to attend the Symposium. Those who have never attended a MSUD Symposium will have priority for assistance although anyone may submit a request. **Funds are limited so don't delay if you are at all interested.** If traveling from another country, the process to obtain a visa and make flight arrangements must begin promptly. Late applications do not allow enough time to obtain visas and make airline reservations. We want to make it possible for everyone to be there who wish to attend. Contact Wayne Brubacher at 574-862-2992 or e-mail: wjbrubacher@afo.net.

A bus from the Lancaster County area of Pennsylvania will be available for those attending the Symposium. To schedule a ride, contact Ivan and Mary Kathryn Martin at 717-354-7732 or email: maryk@hydrosoft.net.

New Website Up and Running... check it out! See article, page 7



Making the Decision to Transplant: Our Family's Perspective

By Sheryl Leinbach

Our family first learned about MSUD when our daughter, Louisa, was 6 days old. The day before, she had been admitted to Hershey Medical Center because she refused to nurse or to respond to us in any way, except for episodes of shrill crying and arching her back. We were so puzzled. What was wrong with our baby who had appeared so perfect and beautiful at birth? (As I had held her on my chest just minutes after birth, she had mesmerized us all by lifting her head and focusing on our faces and smiling at her Daddy's voice.) The doctors did not recognize the symptoms and were doing all kinds of tests to find the problem. Then the PA Newborn Screening program contacted the hospital with a preliminary diagnosis of MSUD. We were so thankful for the Newborn Screening that finally pinpointed the problem.

The hospital transferred Louisa that same day to Lancaster General Hospital to be under the care of Dr. Holmes Morton and Dr. Kevin Strauss, of the Clinic for Special Children. We soon received a crash course on MSUD and hope for a bright future for our daughter. We listened to stories of the first children who were diagnosed with MSUD – children who were brain-damaged and handicapped or who had died during childhood illnesses. We were amazed at the difference Dr. Morton's work had made in the lives of the MSUD children under his care. The technicalities of managing MSUD intrigued us. When we learned that our daughter's outcome depended on the kind of care she would receive, we determined to do our very best. We felt privileged to live close to the most experienced doctors in the world for managing MSUD. Louisa responded very well to treatment, and in five days we took home a baby who appeared perfectly normal again. True, she needed regular blood work and special formula, but she was bright-eyed and smiling, and that was what really mattered.

I remember distinctly that, during one of our lengthy MSUD discussions in Louisa's hospital room, Dr. Morton remarked casually that there is now a cure for MSUD. "There is?! What is it?!" If there was a cure, we certainly would want to do it. Our hopes fell when he replied that the cure is liver transplantation and that he did not really recommend it for us. At that time (summer of 2004), elective liver transplant for MSUD was relatively new. The doctors knew it cured MSUD, but was it a viable and realistic option for those with access to good care for MSUD? We dismissed the thought. It was interesting to know how it worked, but it was too frightening, too huge, too unknown for us to consider.

During her first year, Louisa flourished under the care of the Clinic for Special Children. She was the happiest baby our friends and family had ever seen. She grew and developed in all the normal ways. Her speech and fine motor skills were advanced for her age. We marveled at her every day. "She has a serious genetic disorder, and just look at her!"

The second year she continued to do well, but we began to realize just how difficult it is to control MSUD. She seemed to be very sensitive to even mildly elevated leucine levels and would become ataxic at 5-7 mg/dl. As she became older, she hallucinated very easily. The home DNPH tests were not at all reliable for her. Sometimes they would be clear even though her leucine level was 9 mg/dl. Many were the trips we made to Clinic to check her amino acid levels. They were always available for us any time of the day, night, or weekend to run a complete amino acid test in their lab, and have results within 30 minutes. At 18 months old, she was hospitalized, for the first time since birth, with Rotavirus. Her leucine level was 10 mg/dl, and she was very dehydrated from vomiting and diarrhea. She recovered very quickly and was discharged in four days, completely back to normal. We were so grateful to Dr. Strauss who managed her hospital stay with the expertise necessary to give our normal child back to us. We wondered why anyone would put their child through a liver transplant. Wouldn't it be much easier to move close to the Clinic for Special Children?

At not quite 2 years old, Louisa was hospitalized again with a leucine level of 17 mg/dl. We thought she was recovering from a stomach virus, when her leucine level spiraled out of control. The whole experience was scary for us and left us shaken in spite of the fact that she needed to be hospitalized only a little over 48 hours and seemed to be her perfectly normal self after a good night of sleep in her own bed. We realized just how serious common childhood illnesses can be. Several months later, we were still discussing that hospital experience with the clinic doctors. I asked Dr. Strauss what we can learn and what I had

(*Decision to Transplant* cont. on page 5)

(*Decision to Transplant* from page 4)

done wrong that she needed to be hospitalized with just a common virus. I did not want ever to repeat that horrible experience. I kept reliving the desperate attempts to get formula into a child who was vomiting and our conclusion that her leucine level must not be that high because she wasn't ataxic or hallucinating. I wished we had gone to the hospital sooner… Dr. Strauss said, however, that her hospitalization was not because of a mistake I had made, but because of the nature of MSUD. "MSUD is unpredictable and uncontrollable. We are humbled by the disease." That was not what I wanted to hear! I wanted steps 1, 2, and 3 for the next time. I wanted to be able to control her MSUD.

Wesley and I were both trying so-o-o hard to give Louisa the best! I lived with the constant fear that, in spite of our very best efforts, she could die or be brain-damaged. Dr. Strauss suggested that liver transplantation may be something we should consider, to eliminate the risks and stresses of MSUD. He explained that the fluctuating leucine levels affect the availability of other amino acids which the brain uses to produce neurotransmitters. (We well understood the constant fluctuation of the BCAA levels in spite of our best efforts. We checked her amino acid levels nearly every week, adjusting her diet and formula accordingly.) These imbalances have both immediate and cumulative results, including hyperactivity, irritability, difficulty concentrating, and childhood depression. Dr. Strauss invited us to a Liver Transplant Symposium in Strasburg, to hear from the Pittsburg Transplant Team as well as the Clinic for Special Children.

We were already beginning to understand why other families were choosing liver transplantation. Was it something we should consider? We decided we wanted to learn all we could. Louisa was not quite 3 years old when we attended the symposium. We came away thinking about Louisa's future. She was doing well today, but would she be the same tomorrow? We had seen enough during her last hospital stay to realize just how dangerous elevated leucine levels can be. She had obviously had some brain swelling, which could have been devastating. Wesley and I both agreed that if something serious ever happened to her during an MSUD crisis, we would tend to feel it was our fault. We would feel we should have gotten more formula into her, checked her levels sooner, taken her to the hospital sooner… We would wish we had done liver transplant instead of assuming that she would continue to do well with MSUD. While we realized that something could also go wrong during liver transplant, we decided that we would still feel we had done the very best we could for her. As we weighed the pros and cons, we were gradually convinced that liver transplant really would give her the best future. We thought about school and the difficulty we knew she would have with concentration on days that her BCAA levels were imbalanced. It was inevitable no matter how carefully we tried to control her MSUD. Even though Louisa was not inclined to hyperactivity and had a well developed attention span, school still seemed like a huge thing with MSUD. Our minds could hardly stretch far enough to imagine teenage and adult life with MSUD. We understood that the diet becomes more restrictive after the body is no longer growing and does not need as much protein. A diet consisting largely of artificial low-protein foods could not supply all the necessary micro-nutrients and phytochemicals available from real foods, even if it included the best of formula. If no other options had been available, we would have had hope that she could live with MSUD, accepting it and taking responsibility for her own care in the future. However, since transplant was an option we needed to consider it and make a decision.

Even after we realized that a liver transplant seemed to be the best option for Louisa's future, we still had many questions. What really was life like after transplant? Would we just be trading one set of problems for another? We talked with other families who had done it. We wondered about the anti-rejection medications and their side affects. Medications scare me, so this was a difficult one for me to work through. I needed to evaluate the daily stresses of living with MSUD and weigh them against what I could learn about life after transplant. I did finally conclude that life-long medication is easier than formula, fluctuating amino acid levels, and the "brain" risks during illnesses. And most MSUD patients do need to take medications sometime. Wesley found it easy to look at the facts and make the decision quickly. I was stalled with doubts and fears… To choose liver transplant was choosing trauma. I had to be sure it would be worth it to put my child through something like that. We really appreciated Dr. Strauss's advice. I quote his exact words, "Whatever you decide, be certain that you are unified and committed in your decision. Either path, continued dietary therapy or liver transplant, is likely to present trying times ahead. During such times you will naturally question your decision, and will need each other's support to endure."

Another big question in our minds was the cost of transplant. Since we, for religious reasons, do not carry insurance, where would the money come from? We are part of a strong church brotherhood who believes in helping each other in times of crisis. Would it be right for us to choose to transplant and depend on others to pay the bills? Wesley gave several talks about the decision we were facing to get input and advice from our family and church. We heard only support for going ahead. And really, an MSUD crisis could cost just as much. And what would we have gained?

Even though Louisa was only 3 at the time of our decision, we did feel it important to explain transplant to her. We explained the two options: keeping on with MSUD – drinking formula, eating special foods, not feeling well when levels were imbalanced; or liver transplant – the doctors

(*Decision to Transplant* cont. on page 6)

(*Decision to Transplant* from page 5)

would cut her belly and take her liver out while she was sleeping, she would need to take medicine for her liver but wouldn't have MSUD anymore. She immediately said she wants a liver transplant. Soon she was giving her teddy bear a liver transplant, because he had been in the hospital with MSUD so many times before.

While we were trying to make our decision, a friend had said that when, and if, the time is right to go ahead, the doors will open. They were. The time seemed to be right. Now, before she faced any more serious illnesses. Now, before she was battling with school. Now, while her brain was still young and developing.

As if to clinch our decision, Louisa needed to be hospitalized again. She was vomiting so badly that she gagged if anything touched her tongue. A trip to the clinic showed her leucine at 9 mg/dl. Since there did not seem to be any hope of getting it down with formula we went straight to the hospital. She was on IV for about 6 hours when her temperature suddenly spiked, and she complained of pain in her right side. The nurse called Dr. Morton, and he came to the hospital immediately. Within an hour, she was in surgery for appendicitis. She recovered rapidly and was discharged in less than 48 hours. We were so grateful to Dr. Morton who realized the urgency of immediate appendectomy, which prevented a ruptured appendix and a dangerous infection. We called Pittsburg that very week to start the process for a transplant evaluation. Louisa later had another hospital stay for strep throat, making 5 MSUD hospital experiences before she was five years old. She also had several other stressful illnesses that we managed at home.

At the liver transplant evaluation, we learned details of the actual transplant procedure. This was a major step in putting my fears to rest. The mind views unknowns as huge. I had seen the huge belly scars of other transplantees. All I could picture was the knife. It seemed like such a horrible thing to choose for my child! Yet as the steps of the actual surgery were explained, and we were shown exactly how and where the liver is attached, it did not look so huge. It really sounded so simple, especially from a team who calls it an everyday procedure. We were shown pictures of how transplant patients looked right after surgery, and again it looked like an overwhelming tangle of tubes and wires. Yet when everything was explained, the unknown was removed and we felt prepared and reassured.

We waited 8 months after being listed with UNOS. Louisa was very excited about the prospect of getting a new liver and went smiling into the OR on May 29, 2009. Even though she had a very difficult recovery, spending 7 of the first 9 weeks in the hospital, we in no way regret our decision.

During that difficult time, we clung to the hope that someday we would look back and say it was worth it all. And we do. Louisa has done very well ever since we came home 2 months after transplant. She is so happy to have a new liver. She was able to donate her liver to a boy who had liver cancer, and our families will be connected for life. She has a hearty appetite for all her new foods. She loves the freedom from formula. For myself, I cannot describe the feeling of no worries about amino acid levels! Her care truly is easier than we have ever known. The stress has finally lifted. Even though she may face rejection or other issues, we know it will not compare with MSUD illnesses. We are grateful to the wonderful transplant team at Children's Hospital of Pittsburgh. Most of all, we are grateful to God who has guided and blessed our lives so richly through all our experiences with Louisa, working out all for good.

I could write so much more, especially about our actual experiences. Please feel free to contact us. We will gladly answer anyone's questions. You can call or fax us at (717) 866-6921. ■

Wesley and Sheryl Leinbach and family
Rachel (8), Louisa (5), Ruby (2)

A Few Words From the Editor

Karen Dolins, EdD, RD, CDN
Newsletter Editor

I'm always on the lookout for new research to report on. In December I had the opportunity to attend a conference on the future of newborn screening in Bethesda, Maryland given by the organization Genetic Alliance. This was exciting enough, but it turns out that I got a two-for-one deal. Next door scientists and others were gathered to learn about "Research Challenges in Central Nervous System Manifestations of Inborn Errors of Metabolism." Read the Genetic Alliance article for a brief description of the organization and the conference, and be sure to browse through their website. It has a wealth of information, and teaches us how to advocate to improve the lives of those in need.

As more children are being diagnosed through newborn screening and receiving appropriate care from infancy, they are less affected by the physical and cognitive ailments that have previously been typical of those with this disease. Despite maintaining good control of blood leucine levels, though, many of us have observed that our children struggle emotionally and in school. Unfortunately, there is little to guide us in the scientific literature. Without this support, schools may deny services causing our children to continue to struggle in these domains. Due to my own frustrations in this area, I was thrilled to come upon the article "Screening for Cognitive and Emotional-Social Problems in Individuals with PKU: Tools for Use in the Metabolic Clinic" published in the journal Molecular Genetics and Metabolism in January 2010. While not specific to MSUD, the authors do make recommendations for testing that can be done in the metabolic clinic or in the schools. Even more exciting, the authors, Susan E. Waisbren, PhD and Desirée A. White, PhD at Harvard University and Washington University respectively, have created a network for practitioners in the field to collaborate. Find out more at their website: http://gmpsych.org, and encourage your own practitioners to get involved. Hopefully they will soon publish their experience with MSUD, enabling us to help guide our children through this poorly understood terrain.

We have a vibrant "families" section this issue. You'll all be thrilled to see that we have not 1 but 3 wedding announcements to share with you, as well as other family updates. As you read through the newsletter, please think about ways that you may contribute to future issues.
I look forward to seeing you at the Symposium in June! Find out more about it on page 3. ■

MSUD-SUPPORT.ORG

Eddy Wang (father of Sage, 5 years, classic)
Website Administrator

As of this writing, msud-support.org has 66 registered members worldwide.

As parents and patients of MSUD, we know that communication is key to better treatment and help for those in need. Our goal is to make better use of the tools available on the internet and other new media to better serve the MSUD community. The website strives to bring the informed and non-informed together into a single place to share ideas, feedback, comments and general knowledge about MSUD, a disease so rare that specific information is lacking on established websites. Contributions from our members fill that gap, and our publications will bring an understanding of MSUD to a greater audience.

The msud-support.org website was re-designed in August 2009 to better serve the MSUD community. Many enhancements were made to allow members to contribute their thoughts and ideas. Enhancements include:

■ Finding information using search engines such as yahoo, google, msn and the ability to scour through newsletter articles using a search tool within our website (top right).
■ Member driven content which you can edit. Content must be approved.
■ Question and answer forum – a members only area where registered users are free to post questions and answers.
■ Events posting – fundraising, gatherings, meetings and more.
■ Professionals' directory allowing those in need to find the right doctor or other MSUD specialist (dietitian, physician, geneticist).
Other noteworthy features:
- Donations can now be made online. We use 'Paypal' as our payment gateway which also allows non-paypal users to pay with their credit card/checks.
- Take a survey by voting in our polls. Each month, we will feature a new question for members and non-members to get feedback on current hot topics.
- Directory of MSUD related companies/groups. In this area, you'll find more information about MSUD related products, organizations, companies and friends of MSUD Family Support Group. If you know a company/product/organization you'd like to promote, please become a member to start posting.
- A new video area will be coming soon. It will feature MSUD related videos from all over the internet summarized into categories.

If you would like to see something we haven't thought of, please contact the administrator at admin@msud-support.org.
We hope to see you as members soon. ■

Amy & Randy

Carl and Marty Zimmerman of Lititz, Pennsylvania announce the engagement of their daughter Amy Renee to Randy Gingrich, son of Nelson and Yvonne Gingrich, of Manheim, Pennsylvania.

Amy was born with classic MSUD on September 13, 1984. She had a liver transplant at Children's Hospital of Pittsburgh on January 23, 2006. Amy graduated from Warwick High School in 2003 and Consolidated School of Business in 2004. She works as a medical receptionist at Welsh Mountain Medical Center in New Holland.

Randy graduated from Lancaster Mennonite High School in 2004. He works as a truck driver for Four Seasons Produce in Denver, Pennsylvania. Randy is a two-time leukemia survivor.

A June 12, 2010 wedding is planned.

Jimena Graduates Kindergarten!

In Argentina, when a child finishes kindergarten there´s a celebration for this (they call them "Egresaditos"). Next year Jimena, who will be 6 years old in February, will start primary school. Jimena is doing very well after almost 3 and a half years post-transplant. Happy New Year for everybody!!! Eduardo Gatica, Father of Jimena

My Wedding

By Ruth Nolt

September 17, 2009 was the day Lester Nolt and I exchanged wedding vows. We were glad for beautiful weather as my mother's house was quite full with almost 200 guests.

My levels were just great over the wedding. I must give credit to my parents, Ivan (deceased) and Katie Fox who taught me from childhood to follow my diet closely and to drink all of my formula every day. Without that I couldn't have done all I did and still do!

I have always enjoyed cooking and baking so that wasn't too much of a challenge after marriage. I have learned to make a variety of dishes we can both enjoy or I can make two similar dishes.

I am blessed with a patient and understanding one-of-a-kind husband. It isn't unusual to see him helping me in the kitchen, mixing formula etc. Neither is it unusual for me to ask him how to do this or that in my diet.

I am truly blessed!

Rachel & Kevin

Rachel Marie Wenger of Ephrata, PA and Kevin Lamar Martin of Lititz, PA, both 20, plan to marry on June 5, 2010. Rachel has been on the transplant list for 2 years, and hopes to be transplanted before the wedding. Kevin is cousin to Crystal Martin, daughter of Mark and Lorraine, and part of the MSUD community.

Galen Carrington

is 19 yrs old and attends Wilmington College of Ohio. He has classic MSUD and is currently on the liver transplant list. He is a swimmer for Wilmington Quakers and is the 2nd fastest butterflier on the team. His coach Trip Breen has been very supportive and understanding of Galen's condition and his diet. You can visit the Wilmington College of OH's website, click on Athletics and then click team sports, under men's swimming.

Please send recipes to
Food News Editor

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Cambrooke Foods – Winter 2010

Winter is a great time for trying new recipes and eating good old-fashioned comfort food. Try using meat alternative products such as **Brookelyn Dogs, Veggie Meatballs and Cheese Filled Veggie Meatballs** in your favorite recipes in place of meat. Used with other low protein foods like tortilla wraps, sauce mixes and pastas, you can create favorites like tacos, **Mac-a-Weenie & Cheese, Sweet and Sour Meatballs or "dogs", lasagna and American Chop Suey**. How about an old-fashioned meat ball sub? These meat alternatives also contain 32 mg of DHA, Omega 3 Fatty Acids per serving so you can feel good about what you are serving and eating!

If you are still stumped for ideas, visit Cambrooke's recipe listing under the "Recipes and Tips" tab on the Cambrooke website home-page for ideas to get you cooking!

Struggling to meet your daily protein needs from formula? Try the convenient single serve **Camino pro**® line of formula products for MSUD. Each single-serve product is complete with vitamins and minerals.

Whether you are returning to diet, struggling with your current formula, or just looking for a change, Camino pro™ will help make the low protein diet enjoyable, flexible, and fun. Contact Cambrooke Foods for a sample, to consult with our metabolic dietician.

Request your free Camino pro® sample or place your order today. Call toll-free, (866) 4 LOW PRO / (866) 456-9776 or visit our website at www.cambrookefoods.com. If this is not convenient, you can mail (4 Copeland Drive, Ayer, MA 01432) or fax at (978) 443 -1318.

From Glenda's Kitchen!

Pumpkin Whoopie Pies

2 cups brown sugar
1 cup oil
1 1/2 cups cooked pumpkin
3 cups baking mix (Loprofin or PKU Perspectives baking mix)
1 egg yolk
1 teaspoon baking powder
1 teaspoon soda
1/2 teaspoon cloves
1/2 teaspoon ginger
1 teaspoon vanilla
1 1/2 teaspoons cinnamon
1 teaspoon salt
1/4 cup mini semi sweet chocolate chips

Beat egg yolk, sugar and oil. Add pumpkin and dry ingredients. Drop by tablespoon onto cookie sheet. Bake at 350 degrees for 10-12 minutes or until the top springs back when touched.

Frosting
1 1/2 cup Crisco
3 cups confectionary sugar
1 teaspoon vanilla
1 /3 cup water

Beat together until creamy. Turn half of the cookies upside down and put a scoop of icing on. Top with another cookie sandwiching the icing between the two cookies. 30 Whoopie Pies

Per whoopie pie

Leucine	Protein	Calories
24	.4grams	307

These are very soft, delicious and freeze very well. I have used PKU Perspectives baking mix with great success, too.

Zucchini Fritters

3 cups shredded zucchini
1 teaspoon salt
1 cup carrots grated
2 tablespoons onions
1/2 cup wheat starch
1/8 teaspoon pepper

Mix salt and zucchini and let drain 15 minutes. Press water out. Mix with remaining ingredients. Make into small patties and fry in oil. Serve. 16 fritters

	Leucine	Protein	Calories
Per serving	22mg	.06grams	75

Cheeseburger Soup

3/4 cup chopped onions
1 cup shredded carrots
3/4 cup chopped celery
4 cups diced potatoes
3 cup water
1 teaspoon basil
1 teaspoon parsley flakes
2 tablespoons Major Beef Bouillon

Cook all ingredients together until soft.

White sauce:
4 tablespoons butter
1 1/2 cups water
3/4 non dairy creamer
3/4 teaspoon salt
1/4 teaspoon pepper
1 tablespoon sour cream and onion powder
1/3 cup PKU Perspectives Cheese Sauce mix
Garlic powder

Melt butter and add water mixed with creamer. Add seasonings. With a whisk add the cheese sauce mix and whisk until smooth. Bring to a boil and cook until thickened. Add to vegetable mixture and heat thoroughly. Serve. 9-1 cup servings

	Leucine	Protein	Calories
Per serving	105mg	1.4grams	126

Crunchy Ice Cream Dessert

3/4 cup brown sugar
1/4 cup melted butter
2 1/4 cups Ener-G Foods low protein pretzels (crushed)
3 cups Cool Whip, thawed
1 small package instant vanilla pudding
Strawberry ice cream topping

Combine sugar, butter in a sauce pan. Heat until sugar is melted. Mix with pretzels. Press 1/2 of the mixture in a 9 x 13 pan. Put into refrigerator until firm. Mix Cool Whip and instant vanilla pudding together. Spread over pretzel crust. Top with remaining crumbs. Freeze until firm. Drizzle with Strawberry ice cream topping when ready to serve. 12 servings

	Leucine	Protein	Calories
Per serving	36mg	.5grams	198

Genetic Alliance transforms health through genetics and its network includes more than 1,000 disease-specific advocacy organizations as well as thousands of universities, private companies, government agencies, and public policy organizations. The network is a dynamic and growing open space for shared resources, creative tools, and innovative programs. Through its work, Genetic Alliance brings together diverse stakeholders to create novel partnerships in advocacy, integrates individual, family, and community perspectives to improve health systems and revolutionizes access to information to enable translation of research into services and individualized decision-making.

On December 7 - 8, 2009 Genetic Alliance hosted the **Newborn Screening Summit: Envisioning a Future for Newborn Screening** in Bethesda, MD. More than 200 health professionals, family members, and others participated in the summit to discuss how long-standing state public health programs can evolve to keep up with new technology, societal and governmental trends, and medical advances. "This meeting provided a wonderful opportunity to visualize the newborn screening system from the point of view of many stakeholders. Working together we can provide the support the entire system needs," said Sharon Terry, President and CEO of Genetic Alliance. The meeting was webcast live, and the videotaped proceedings as well as more information can be accessed at: http://www.geneticalliance.org/ws_display.asp?filter=nbs.summit.

RESEARCH CHALLENGES IN CENTRAL NERVOUS SYSTEM MANIFESTATIONS OF INBORN ERRORS OF METABOLISM

Department of Health and Human Services, National Institutes of Health's Office of Rare Diseases, the National Institute of Neurological Disorders and Stroke, FDA Center for Drug Evaluation and Research

Concurrent with the workshop on Newborn Screening, this meeting took place in an adjacent room. The purpose of the meeting was to review and discuss the difficulties in translating preclinical studies in animals to clinical trials in humans.

Individuals with IEM have a range of functioning, which may vary with the specific mutation. The neurological effects of disease over time are virtually unknown. The presenters noted that researching inborn errors of metabolism (IEM) is made particularly challenging by their rarity, ethical issues in studying children, and the fact that it is extremely difficult to study the impact of disease and its treatment on the brain. What is clear is that IEM, including MSUD, are diseases which affect the nervous system. As such, the individual may not be functioning optimally even when well controlled.

The opening speaker noted that while attempts to treat IEM with stem cell transplant or enzyme enhancement have been unsatisfactory to date, advances in biology and technical tools used to manipulate cells have occurred.

Lay organizations such as ours are invaluable to scientists as parents and patients teach the doctors, conduct fundraising to support research, and act as advocates to legislators. The speaker noted that organizations which raise money for research must have a scientific advisory board to review grant applications. An independent assessment is needed to evaluate the scientific credibility of the proposed research and to balance the risk with potential benefit.

The room was filled with physicians, researchers, public health officials, psychologists, genetic counselors, representatives of support groups, pharmacologists, and more. As I observed all these individuals, organizations, and medical institutions speak of the need to collaborate, I felt a real hope that a breakthrough will occur. ■

HELP WANTED

Are you up for a rewarding and stimulating volunteer job? I'm looking for a co-editor of the newsletter. The time commitment is about 10 hours twice a year, and involves reaching out to potential contributors and reviewing articles as they come in. Please let me know if you are interested. It's a great way to connect with the MSUD community. Please contact me at krdhed@aol.com Thanks! Karen Dolins, Newletter Editor

Our Tryst with Destiny

By Prashant Verma

Shourya and Mom, Niti

Shrill cries, semi-comatose condition, feed rejection & hospital as first home, is something which many of us may have encountered over the course of our lives with MSUD. Our story is not different from many of yours as we welcomed our first child along with the experience of MSUD. Our story though is slightly different from many as the country that I belong to is the 2nd most populous country of the world, has the highest population growth in sheer numbers, yet medical amenities are basic and the care for metabolic disorders is a far cry from countries in the West. Just to paint a scenario, India is roughly six times in population size in comparison to USA and has the highest child population in the world. Even though disorders associated with birth is quiet large in number, there is next to nil knowledge or expertise available in the arena of metabolic disorders and in-born errors of metabolism.

Shourya was born on March 1, 2007. With all our wishes coming true and post a normal delivery he was back home the very next day to be a part of the family in person. However the destiny had already decided something else for us and on the 7th night after his birth, he was back in the hospital on a ventilator battling for his life. The doctors tried every permutation & combination including a change in hospital but all in vain as nearly all tests suggested normalcy. The doctors were completely confused and perplexed. On the morning of the 9th day, as Shourya's condition was deteriorating, one of the doctors suggested a metabolic disorder. On the 10th day evening it was confirmed that he is suffering from MSUD. The only thing the doctors could tell us was to read more on MSUD on the Internet as they themselves had never encountered a patient of MSUD. And some so called well-wisher doctors advised us to abandon him as survival of such cases is an issue beyond a month or so, and on top of it feed (*editor's note: metabolic formula*) is not available in India and getting it from abroad will make us go bankrupt in a few months.

Destiny possibly was testing us and we decided that be hell or high waters, we will give our best shot. With the help of internet, we got in touch with doctors abroad and procured some feed which was delivered to us at a lightening speed. From the 12th day onwards Shourya was on the MSUD diet and started showing signs of recovery. In between, Shourya underwent peritoneal dialysis. The internet and tele-communication played a huge role in saving the life of Shourya. And with the help of doctors abroad & locally, we started managing Shourya on a daily basis without even having an inkling of what the future entails for us. We just believed that the Almighty is there with us & as told by someone that special kids come to some chosen ones, we were very clear that Shourya will lead a normal life with us.

In the next few months, we took him to the Great Ormand Street Hospital (GOSH) in London and that was our first lesson with managing MSUD on a daily basis. The doctors there helped us to understand the feed management, the daily assessment of kids and management of the sick day regime. Shourya was by now 3 months old but he was delayed as most of his time was spent in hospital & his nutrition & feed was not managed properly, thus making a baby who weighed lesser than his birth weight by this time now. We realized that MSUD indeed is not as difficult & tragic as it was explained to us initially. But, for us the challenge really was that back in India, there were no doctors who could take care of him in emergency situations. Above this in India there are no feed manufacturers & hence the entire stuff had to be imported always either from US or UK without any financial help from the Government or the Insurance companies.

For the next 2 years, we kept importing the feed and every time we went to London & USA for his regular review, we ensured that we stack up our luggage with his feed, and that we wouldn't fall short of his daily requirements. Not only the cost of these medicines are huge, which we all know but, to import the same in my country there is 200% import duty and thus typically one tin of Analog or MSUD complex which costs say $50 in USA, the landed price for us including courier was somewhere around $180-$190.

We ensured that Shourya never misses his weekly HPLC test & twice a week DNPH tests. We tried to insulate him from normal infections as much as possible, but going forward we knew that it will be a tough ask to manage him like this for long as:

1. With Shourya growing up, his feed requirements were increasing and with no variety available continuing this scenario for long was becoming a challenge.
2. With Shourya growing up & by the 15th month he was on track with all his developmental milestones, it was a tough task to keep him away from normal food and children at large….

(*Shourya* cont. on page 13)

Update on our Work with a **Mouse Model** for Understanding MSUD

Dr. Susan Hutson

A major goal of our research is to understand the molecular basis for the effects of MSUD on the brain as a means to developing effective therapy.

We have generated a mouse line that has what we call a floxed E1-alpha gene that will allow us to ask very fundamental questions about MSUD and brain pathology in an animal model. The animals that we have generated (floxed E1-alpha) have DNA pieces put into them that are recognized by the enzyme Cre-recombinase. By breeding with what we call a Cre-mouse (a mouse which expresses the Cre-recombinase enzyme), we are able to remove a portion of the E1-alpha gene and produce an animal with a defective E1-alpha gene. Through several breeding cycles, we first produce animals that have the defective gene (deletion) in one chromosome (+/- animal) then breed those to make animals that have the deletion in both chromosomes (-/- animal). This is the knockout or KO (-/-) animal, because this animal does not produce the E1- alpha protein (or produces a defective protein with a large piece missing). Today there are mouse lines that permit us to create a knockout in the entire animal or in specific organs and tissues. Ultimately we want to generate a mouse that does not express E1- alpha only in the brain.

We have generated MSUD E1 knockout animals by breeding mice heterozygous (one chromosome has the deletion and the other one is normal) for the E1-alpha gene deletion in both chromosomes (-/- or KO animal). These animals are classic MSUD and, as found with the E2 total knockout (*KO), the pups die within 1-2 days of birth. We are using these animals to show the localization of the branched-chain alpha-keto complex (BCKDC) in the brains of the mice. The enzyme is located in nerves (neurons) and not in other types of brain cells, but expression is not uniform throughout the brain. Right now we are breeding the neuron-specific Cre mouse to establish the line here, and then we will mate them with our E1 animals (floxed E1). Animal breeding is a time-consuming process. We hope to produce the desired animals in the next couple of months. The phenotype of these animals will show whether altering brain metabolism is sufficient to produce the neurological effects and determine the next course of experiments. Stay tuned for further updates.

We are also collaborating with Dr. Brendan Lee at Baylor on the phenylbutyrate project. Our laboratory will be doing the cell work, measuring leucine metabolism and the effects of phenylbutyrate on MSUD patient lymphoblast cell lines to determine whether we can predict efficacy.

Editor's Note: Please see Dr. Hutson's article in the Fall 2007 Newsletter for more information on this project.
Editor's Note: Please see Dr. Lee's article on front cover.

(*Shourya* cont. from page 12)

By now, we felt assured that minor infections & emergency regime protocol was not so much so a big problem for us or for the doctors but critical illness management was still a challenge for the doctors because of no experience, lack of TPN availability and general acceptance of MSUD disorder as a priority in a hospital. We had by now, seriously started contemplating a liver transplant for Shourya, though some doctors in UK as well as in India believed that managing Shourya with MSUD is a better option than undergoing for a liver transplant.

I visited the June MSUD symposium at OHIO in 2008 and met up with most of the MSUD kids & adults who underwent the transplant and their parents. I also went and met up the Pittsburg child hospital team and when I returned back to India, I was quiet clear that we will be going for liver transplant. The only question was when and where? We immediately got in touch with the liver transplant unit at Sir Gangaram Hospital in Delhi and the doctors there were very confident that it can be done. They parallely started their research with doctors abroad and within a month, we did a pre transplant evaluation for Shourya. Every possible research that was required, including a DNA assessment was done for the transplant process; with a view to make transplant happen asap. Getting a cadaver organ is next to impossible out here, however in India live donor transplant is quite common and hence we just needed a donor to make it happen.

By this time, the destiny had a different color and my sister-in-law Mukta, immediately came forward to donate her liver. As liver grows back in the donor's body within 1-3 months to its original shape & size this mother of 2 kids took up this great challenge for sheer love & affection for our baby. She was our Lady Nightingale.

Shourya was transplanted last January inheriting 20% of the liver from his aunt Mukta & in turn gifted his liver to Sia, a baby girl of 3 years who was suffering from LCH (Langerham cell Hystocytosis) with a terminal liver failure. The Live donor cum Domino transplant happened for an MSUD case for the 1st time in the world. Shourya is also one of the youngest donors in the world today. Today we are very happy that all the 3 (Mukta, Shourya & Sia) are doing well. Like his Aunt Mukta, Shourya is a proud donor to Sia and we firmly believe today that no condition is non-manageable, yet to manage a condition; one must ensure a thorough discipline.
We are greatly indebted to many doctors and their support staffs both in India and abroad who have worked relentlessly and also my family and friends who not only provided physical, mental and emotional support but also stood by us during this journey. ■

Marlon

was diagnosed with classic MSUD at three days of age, through the newborn screening available to all newborns in the state of Alaska. Before we could notice any of the signs that he was showing, (lethargy, stiffness in his body and lack of appetite), the pediatric office contacted us to inform us of his diagnosis. Marlon was the first newborn ever diagnosed with MSUD in the state of Alaska, so getting the right treatment was difficult. To keep Marlon metabolically stable on the first days of his life we needed the special MSUD formula and right IVs. He was hospitalized right after he was diagnosed with MSUD, but the formula was sent by mistake to a far away city in this same state. It took 2 days for the formula to reach us, and by that time Marlon's leucine levels were higher than 1900. Marlon had to be med evacuated in a jet from Anchorage, Alaska, to Seattle, Washington where he would undergo hemodialysis. He would remain in Children's Hospital of Seattle during 16 days until his amino acid levels become normal and stable.

Marlon, with parents

The initial crisis was the worst that Marlon suffered in the four and a half years that he lived with MSUD. Several crises followed that one due to common childhood viruses, such as the Rota virus. During those crises he was able to receive treatment in the local hospital in Anchorage. He recovered fine from all of these episodes, but we always wondered if there had been neurological damage.

When Marlon was two years and a half we visited Spain, my home country. Marlon started showing severe signs of ataxia. We tried to control the elevated leucine levels by supplementing his MSUD formula with isoleucine and valine. Although we kept him hydrated and drinking his daily formula, the ataxia continued and became each day more severe. He was hospitalized in Spain but MSUD was unknown there and we soon saw ourselves trying desperately to get out of the country to get back to Alaska to seek treatment. I never felt like I could take Marlon back to Spain again safely since the last experience was very frightening for us.

It was around that time when the first liver transplants were done to MSUD children to free them from the symptoms and risks of living with MSUD.

Marlon

fue diagnosticado con MSUD a los tres días de edad a través del test neonatal que se practica a todos los recién nacidos en el estado de Alaska. Antes de que nos diésemos cuenta de los síntomas que presentaba (letargia, rigidez del cuerpo e inapetencia), nos contactaron de la oficina pediátrica para informarnos de que tenía MSUD. El estar en Alaska y el ser el primer niño diagnosticado con esta enfermedad metabólica en el estado dificultó mucho el tratamiento adecuado en los primeros días de vida. Tras ser hospitalizado en nuestra ciudad, la fórmula para MSUD fue enviada por equivocación a una lejana ciudad de Anchorage, también en el estado de Alaska. Dos días pasaron hasta que la fórmula por fin llegó al hospital donde se encontraba nuestro hijo. Cuando la recibimos los niveles de leucina de Marlon superaban los 1900. La crisis metabólica inicial fue la peor que sufrió en los cuatro años y medio que vivió con esta enfermedad. Marlon tuvo que ser evacuado via jet del estado de Alaska al de Washington para poder recibir hemodiálisis. Permanecería en el Children's Hospital de Seattle durante 16 días hasta que sus niveles de aminoácidos se normalizaron. Sufrió varias hospitalizaciones a causa de las crisis metabólicas que se le presentaron como consecuencia de virus comunes infantiles, como el rota virus. Durante estas crisis pudo recibir los cuidados necesarios en el hospital local de Anchroage. De todas las crisis salió bien, pero siempre nos quedábamos con la incertidumbre del no saber a ciencia cierta las secuelas que estas crisis pudieran haber dejado en Marlon.

A los dos años y medio y cuando nos encontrábamos en España visitando a nuestra familia, Marlon comenzó a presentar severos síntomas de ataxia. Intentamos corregir los elevados niveles de lucina suplementando con valina e isoleucina, y a pesar de que ésto nos ayudó a mantenerle con apetito, tomando su fórmula de MSUD, la ataxia no sólo continuaba sino que seguía haciéndose más severa.

Ante el desconocimiento de los tratamientos para las crisis metabólicas de MSUD que había en los hospitales de la región que visitábamos, nos vimos obligados a viajar de regreso a casa, Alaska, semanas antes de lo previsto. Nunca más me sentí con las fuerzas ni el valor de viajar con Marlon fuera del país, ya que la última experiencia había sido sumamente aterradora, y con ésta se truncaba la posibilidad de volver a visitar a mi familia. Por aquel entonces comenzaban a hacerse transplantes de hígado a pacientes con MSUD para librarles de la sintomatología y riesgos de MSUD.

(*Marlon* cont. on page 15)

(*Marlon* cont. from page 14)

When Marlon was about three years old we decided we would look for a surgeon who would be knowledgeable in MSUD and who would be willing to transplant Marlon. Many months went by searching the way to plan a transplant for our son. Many phone calls, petitions for insurance coverage, appeals, and long trips out of the state of Alaska continued for months. Finally we were able to plan a liver transplant evaluation for our son in Seattle Children's Hospital. On February 18th, 2005 Marlon was officially listed to receive a liver with a score of 40 points. We would have to wait 13 more months for a liver transplant. In the early hours of March 30th, 2006 we received the so long hoped for phone call that would announce that there had been a donation that was the perfect match for Marlon. In the next few hours we underwent many emotions, a flight to Seattle, preparations at the hospital, and a meeting with the Chanel King 5 TV team. Marlon went into OR at 12:15 am on March 31. It was a long night, and at 8:30 am we were told Marlon's transplant had been successful and he was heading to the PICU. He would be hospitalized for 19 days. The first two weeks were filled with progress daily, but during the second week he had an episode of rejection that required the appropriate treatments and delayed Marlon's recovery. He responded very well to the treatments and he was released from the hospital on the 19th day post transplant. We would live in a nearby hotel for 6 more weeks to be able to attend clinics and post transplant checkups.

The following months after receiving his new liver Marlon would keep progressing with a normal diet, no more restrictions, no more low protein products or MSUD formula. He was able to start preschool 6 months post transplant. One and a half years post transplant Marlon suffered again from rejection of the liver. We had to travel to Seattle again to be able to give him the appropriate treatments. He responded very well and promptly and once his liver functions were normalized we traveled back home in Alaska.

Nowadays Marlon lives a life free from MSUD. He just turned eight years old, he attends a regular school and he is in second grade. He is learning how to ski on the slopes where his dad learned as a child. He dreams of becoming a surgeon, learning from Dr. Reyes (his transplant surgeon). Until not long ago he would give transplants to all of the super heroes in his collection. He loves to draw, watch movies, he speaks English and Spanish fluently and he is on third year of piano. He loves to travel and visit his family in Spain, where we spend two months every summer to enjoy the Spanish culture, to have some quality time with family, practice the language, and of course, enjoy the Spanish Mediterranean diet, in particular seafood and legumes, which Marlon loves with passion. He now is able to spend hours in the warm and sunny beaches of Spain without suffering from a metabolic crisis, and he is able to stand the many degrees below of the cold winters of Alaska with no consequences.

We are eternally thankful to those who were pioneers on the cure of MSUD through liver transplants, thankful for life to the surgeon and our hero, Dr. Reyes, Chief of Transplant Surgery at University of Washington, Seattle. And most of all thankful to a family who, while they lived a tragedy, showed the biggest gesture of generosity that someone can ever think of, the donor family. ■

Cuando Marlon contaba unos tres años nos dispusimos a buscar un cirujano que estuviese dispuesto a transplantar a nuestro hijo. Siguieron meses de investigación, llamadas telefónicas, peticiones al seguro médico y viajes largos fuera del estado de Alaska, hasta que al fin logramos una evaluación para un transplante de hígado para nuestro hijo en el Children's Hospital en Seattle, estado de Washington. El 18 de febrero del 2005 Marlon entraba oficialmente en lista de espera con una puntuación de 40 (máximo). Siguieron 13 meses de larga espera por el órgano adecuado para nuestro hijo. Y fue en la madrugada del 30 de marzo del 2006 cuando sonó el teléfono para alertarnos de que había una donación pediátrica compatible con Marlon. Siguieron horas de muchas emociones, vuelo desde Alaska a Seattle, preparación en el hospital, equipo de cámaras del canal King 5 y finalmente ingreso en el quirófano a las 12:15 am del 31 de marzo. La noche entera pasaría lentamente hasta que a las 8:30 de la mañana nos dieron la noticia de que el transplante de Marlon había sido un éxito. Siguieron 19 días de hospitalización. Las dos primeras semanas fueron constantes progresos, pero un pequeño rechazo de órgano retrasó la recuperación. Tras recibir los tratamientos adecuados Marlon era dado de alta en el hospital a los 19 días de su transplante. Por 6 semanas permaneceríamos en un hotel en la ciudad de Seattle para atender a las clínicas y revisiones post transplante. Siguieron meses de recuperación, progresos en la dieta, totalmente normal tras recibir su hígado, y disfrutando de una vida sin síntomas de MSUD. Comenzó su escolaridad a los 6 meses del transplante. Al año y medio del transplante Marlon sufrió una elevación en las funciones hepáticas necesitando otra hospitalización para atajar el rechazo. Respondió óptimamente a los tratamientos y volvió a la normalidad.

El día de hoy Marlon vive sin MSUD, acaba de cumplir 8 años, atiende al colegio con niños de su edad, está aprendiendo a esquiar en las pistas de ski donde su padre aprendió de niño, sueña con ser cirujano y dueño de un restaurante en el futuro, practica transplantes en su sala de operaciones a todos los super heroes de su colección. Le encanta dibujar, ver las películas de moda, habla dos idiomas con fluidez, está en tercer año de piano y en segundo grado en su escuela. Le encanta viajar y visitar a su familia española y pasamos cada verano en mi tierra natal para disfrutar de la cultura materna, conservar el idioma y cómo no! para disfrutar de la culinaria de España, la favorita de Marlon, a quien le encantan los mariscos y legumbres de la dieta mediterránea. Puede pasar horas en las calurosas playas de España sin desarrollar una crisis metabólica, o aguantar los muchos grados bajo cero durante los fríos inviernos de Alaska sin sufrir consecuencias metabólicas.

Hoy en día estamos eternamente agradecidos a aquellos que fueron pioneros en investigar la posibilidad de una cura para MSUD a través de un transplante hepático, agradecidos de por vida con el cirujano y nuestro héroe, Dr. Reyes, Jefe del Departamento de Transplantes del Children's Hospital de Seattle, y sobre todo a la familia que en medio de su desgracia tuvo el mayor gesto de generosidad que alguien pueda tener, la familia donante. ■

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NUTRICIA North America
formerly SHS North America

This publication is supported by a grant from Nutricia North America