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MSUD NEWSLETTER

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2010 MSUD SYMPOSIUM
LINCOLNSHIRE, ILLINOIS JUNE 24-26

For those of you who were unable to attend the 15th MSUD Family Support Group Symposium, and for those of you who welcome a reminder, here is a brief synopsis of the lectures and events:

Loren Pina, MD, PhD is a geneticist in the Department of Pediatrics at the University of Illinois Medical Center in Chicago. She provided a historical perspective of MSUD, reviewed the biochemical defect involving the decreased activity of the branched-chain ketoacid dehydrogenase complex which results in elevated levels of leucine, isoleucine, and valine and their keto-acids, and outlined symptoms and diagnostic procedures.

Barbara Burton, MD is Professor of Pediatrics at the Northwestern University Feinberg School of Medicine and an attending physician in the Division of Genetics, Birth Defects and Metabolism at Children's Memorial Hospital in Chicago. Dr. Burton discussed treatment of MSUD, the need to closely monitor diet records and growth of the affected child. Current treatment of metabolic decompensation includes treating the illness or other factor that caused the event, withholding leucine from the diet, and providing adequate calories and other amino acids to promote protein synthesis. This can be accomplished orally, via naso-gastric or gastrostomy tube, or intravenous infusions of glucose, lipids, and BCAA-free TPN. When necessary, hemodialysis can be used to bring down severely elevated leucine levels. Future therapeutic options include new drugs such as biphenyl, hepatocyte transplantation, and gene therapy.

(*Symposium2010* cont. on page 2)

SYMPOSIUM REFLECTIONS
Barbara Mudrick, Symposium Chair

As I finally sat down a few weeks after the symposium to review all the feedback I was just amazed at what an incredible and diverse group we have. Everyone had such wonderful things to say about the speakers and venue location, but most importantly the support in meeting the different families.

Here are come comments made by attendees of the 15th Bi Annual MSUD Symposium:

"The MSUD Symposium was a total breathe of fresh air. I have learned more than I thought feasible. Interacting with other families and the professional speakers was inspirational. Words cannot really explain how much I have learned and how much

(*SymposiumReflections* cont. on page 2)

Inside This Issue:

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better I feel about Ruby's future." – Colin Kirwan, Australia

"I learned that in my lifetime I am going to live to see a cure or at least something that will enhance the quality of life. I have gained hope and inspiration."- Katie Foster

"I came here to learn about having children and the liver transplant. I learned so much and met so many new friends around my age. I am excited to learn more and to keep in touch. Thank you for all the information. It was a great Symposium!!" -Amanda Walton

"Wonderful Symposium! Thanks for all the dedicated work of the planning committee."

"Had a fantastic time for our first symposium. Very informative."- Sarita Garman

Suggestions for future symposium locations include Sydney, Australia, Honolulu ,Hawaii, Boston MA, Columbus OH, Raleigh NC, Lancaster PA, Ann Arbor MI, Atlanta GA, Salt Lake City, UT.

And the winner of the 2012 MSUD Symposium is…… Just kidding- you will have to wait until the spring newsletter to find out, but if you are interested in getting more involved please contact Barbara Mudrick at barbmud@yahoo.com or Sandy Bulcher at dbulcher@aol.com.

When I volunteered to organize this year's symposium, I knew that this was going to be different than putting on a work event, but I never expected to have such a rewarding feeling from doing something like this. When the board came to Chicago to view the Marriott almost 2 years ago there was some hesitation about my commitment to the group since we had decided to put Dylan on the liver transplant list. I assured them that MSUD is part of our lives and will always be. Dylan did have his transplant Aug 8, 2009 and I'm still here. I feel that no matter what a family's personal decision is that you should still support the MSUD community, whether financially or attending a symposium. I want Dylan and his brother Ethan to grow up understanding what an amazing group of people have this disease and to never take life for granted. I can't begin to say how proud I am to be involved with such a great group. ■

Barbara Mudrick, 2010 Symposium Chair

(*Symposium2010* cont. from page 1)

George Mazariegos, MD is the director of Pediatric Transplantation at Children's Hospital of Pittsburgh. He updated us on the status of liver transplantation for MSUD, noting that while this procedure was historically limited to life-saving situations, it is now considered an option for improving life. Liver transplant provides a metabolic cure for MSUD, allowing the removal of dietary restrictions and near complete protection from decompensation during illness. These benefits must be weighed against the risk of surgery, rejection, immunosuppression and infections

Jeff Masse is a personal chef in Milwaukee, Wisconsin. He provided an entertaining cooking demonstration and allowed participants to taste delicious low protein foods made with all natural ingredients. Highlights included a watermelon gazpacho, minestrone salad, French potato salad, and confetti coleslaw.

John Parker, a motivational speaker, brought his life stories to the group. ■

Evidence-based Treatment Plans for MSUD

Dianne M. Frazier, PhD, MPH, RD
University of North Carolina
Chapel Hill, NC 27599

With the expansion of newborn screening, it is now conceivable that every infant with MSUD can be detected in the newborn period, and treatment initiated to allow them to have healthy and productive lives.

With earlier detection comes a need for evidence-based treatment guidelines. Those involved in newborn screening programs have devoted much of their energy over the last decade to the development of newer laboratory methods for detecting numerous rare inborn errors of metabolism (including MSUD). Some of the disorders that can now be detected are so rare that there has been very little accumulated data about best means of treatment. Furthermore, the treatment protocols for most inborn errors of metabolism are as variable and numerous as the clinics providing care. With funding from the federal agency Health Resources and Services Administration (HRSA), the Genetic Metabolic Dietitians International (GMDI) and the Southeastern Regional (SERC) Collaborative have undertaken a project to create evidence-based nutritional treatment guidelines for inborn errors of metabolism detected through expanded newborn screening.

The nutritional guidelines project expects to achieve the following:

- Less uncertainty and variability in the management of patients
- More consistent documentation of care and tracking of outcomes
- Ability to combine clinical data from many centers to inform and evaluate practice
- Identification of priority questions for collaborative research

A group of eight senior metabolic dietitians from across the US make up the workgroup devoted to writing the guidelines for MSUD. They have identified 5 topic areas on which to concentrate their efforts:

- Use of thiamine supplementation
- Treatment during pregnancy and the postpartum period
- Treatment during acute illness and at diagnosis
- Treatment during and after liver transplantation
- Optimal plasma levels of branched chain amino acids

The process being utilized for gathering data is called evidence-based analysis. This is a standardized process by which data from published research and clinical articles are evaluated and combined to see if there is strong evidence to support various aspects of treatment. Because many clinics have developed unique treatment protocols based on their own experience with their patients with MSUD, a collection of these clinic protocols is also being evaluated. In instances where all the accumulated data fails to provide sufficient evidence to allow for writing a clear guideline, the workgroup has developed a survey tool to be taken by experienced metabolic physicians and dietitians from all regions of the US. Data from the completed surveys will be combined, and any items that remain unclear will be discussed in a face-to-face meeting of metabolic experts. A concensus will be reached based on scientific reasoning. We will ask for input from the MSUD community before finalizing the guidelines.

What will the final guidelines look like? They will contain background information about MSUD, its diagnosis, biochemistry and genetic characteristics, all adequately referenced. Next, step by step nutritional management guidelines will be given to cover all aspects of care and monitoring from the time of diagnosis, during acute illness, and special circumstances over the lifespan. Each step of the guidelines will have annotations informing the user of the strength of the evidence supporting that particular step. Finally, there will be resources and links to resources that will be helpful for managing MSUD.

While the guidelines are being developed for the healthcare team, their ultimate goal will be to provide the best possible care for the MSUD community.

Should you wish to participate in the public input, you may contact me at dfrazier@med.unc.edu ■

Ruby Maree Kirwan

1 Year Old

By Colin Kirwan

The 25th May 2009 at 17:07hrs. This is the day our lives would change forever and in more ways than we could have ever imagined. Our beautiful little girl weighed 3.61kg (7.95 pounds) and both Mum and Ruby Maree were doing fine.

We were home by Day 2 and the community nurse called on Day 3 to check how things were going. Normally this is when the heal prick test would be done but she had run out of time and said "not to worry these things never come back positive for anything" so we'll try make time tomorrow. The blood sample was taken on Day 5, and was not immediately sent to the lab resulting in a further delay of the test.

Everything was generally good for the first few days. Ruby was a fussy feeder and Fiona was attending various breast feeding classes to try and overcome any problems in this area. On around day 7 or 8 we were becoming more concerned about Ruby's lack of appetite. We had been referred to a breast feeding specialist who was concerned at Ruby's back arching and the weird cry she was making. Ruby was referred to a chiropractor! to evaluate the back arching problem. On Day 10 Fiona took Ruby to the Chiropractor who believed he has found an issue with her back and booked further appointments.

I phoned Fiona to see how it was all going. She told me that Ruby had not eaten since that morning so we agreed to meet at the community nurses office for her scheduled checked up. Now anyone who knows me will suggest that I can be a bit of a drama queen, but when I saw Ruby I just knew something wasn't right. Our community nurse also seemed very concerned but was not able to offer a reasonable explanation. Fiona and I both agreed that a trip to the hospital was now required. The local hospital was expecting us when we arrived. At the same time my mobile phone rang. Being in a highly stressed state, that phone was almost vaporised by my glare of anger at who could possibly be calling at this moment. I thrust the phone at Fiona for her to deal with as I doubted I could answer it without being very rude.

Whilst triage was doing the things they do I noticed that Fiona was still talking on the phone. I suggested she tell them to politely go away. This is when I noticed Fiona had gone white and was quite upset. The person on the other end of the phone was a metabolic doctor from the Royal Children's Hospital and she had bad news. They had been trying to get a hold of us by calling area hospitals and had even sent a doctor to our home. They didn't tell us what was wrong with Ruby, only that she was very sick and that they were sending an ambulance. The doctors at the local hospital were reluctant to answer any of our questions as they had never heard of MSUD. Ruby's diagnosis came

on Day 10 due to the failure to follow the correct protocol for newborn screening. They should have been able to tell us 6 days earlier.

A paediatric life support ambulance was called. Things where happening very quickly now and our nightmare had commenced. Our little Angel had become encephalopathic and was not responding. She was rushed to intensive care and was placed on coma inducing drugs and Haemofiltration. We were not allowed to touch or stimulate her as they wanted her brain to remain as unstimulated as possible.

We were taken to a room filled with people: metabolic section heads, fellows, dietitians and social workers. It was almost midnight and we were ready to explode. "Your daughter has MSUD she probably has brain damage but we won't know to what extent for some time" is all I remember from that meeting.

Ruby spent two weeks in Intensive Care. She had an MRI which showed "extensive edema in the myelinating pathways" or damage to her brain and had also ended up with a large clot in her leg from the haemofiltration.

We spent the next 12 weeks giving our little baby 2 injections a day of clexane to thin her blood. They even got this wrong. We received a phone call telling us that "Ruby has been

(*RubyMaree* cont. on page 5)

Dietary Treatment of MSUD

By Heather Bausell, RD, LDN
Children's Memorial Hospital

Registered Dietitian Heather Bausell taught attendees the concept of using the MSUD Food Pyramid. Components of the pyramid include medical formula, fruits and vegetables, low protein foods and starches, and fats, oils, and free foods. She noted the importance of providing adequate calories to prevent catabolism (tissue breakdown), and the need to supplement isoleucine and valine so adequate growth can occur.

Protein is as important to those with MSUD as it is to those without. The trick is to get the right amount to support growth without too much of the branched-chain amino acids (protein is made up of amino acids). Inadequate protein will result in impaired growth along with more frequent infections as the body lacks the necessary antibodies (which are proteins) to adequately protect against them.

Heather provided tips for making formula creatively, including adding fruit for a smoothie, mixing it with juice, Gatorade, or coffee, or even adding it to salsa, ketchup or applesauce!

Essential fats are needed for a healthy brain, nervous system, vision, and healthy skin. Oils provide fats as omega-3 or omega-6 fatty acids. Omega-3 sources, including flax oil, walnut oil and canola oil are particularly helpful in fighting heart disease. They can be used to cook vegetables or pasta, as a dip for bread, and as salad dressing.

Heather concluded her talk with a case study of a young boy who had frequent hospitalizations and difficulties taking in adequate formula. Concerned about his health and the frequent battles surrounding feeding, his family elected to have a gastrostomy ("G") tube placed. Family battles were gone and illnesses were able to be managed at home without hospitalizations. ■

"Life is nothing until it is lived, but it is yours to make sense of; the value of life is nothing other than the sense you choose."

-Jean Paul Sartre

(*RubyMaree* cont. from page 4)

receiving the wrong dose please don't drop her or let her fall she could bleed internally!!"

We understand there are less than 12 MSUD people in Australia. We have never met any of them. Our system is not conducive to family support. I located a group through Face Book and more importantly a lady known as Barbara Mudrick. "Why don't you come to America we're having a symposium you could learn a lot". My reply "geez thanks for the offer but it simply isn't possible. The last 9 months have drained all our resources." Barbara's reply, "leave it with me I know a group that may be able to help."

Fast forward: now Ruby is 12 months old and has had her fair share of complications and hospital visits. Barbara has been true to her word. Through the MSUD family support group, Wayne and Joyce Brubacher and a private backer "Janine", I'm on an aeroplane and on my way to Chicago for my first Symposium.

My first big trip, 20hrs of plane travel, wow! Who are these people? The symposium was simply the best thing that has happened for our family in the last 12 months. In addition to our battle with MSUD I had been laid off my job 4 weeks prior to departure, but we pushed through and here I was in Chicago! I met a lot of people at the symposium and I simply can't mention them all but you know who you are. I asked lots of questions that only you guys could answer. Diet, "G" tubes, education, social interactions, babies, the list goes on. I think the best part of this was meeting people with MSUD and knowing that medicine today is leaps and bounds better than 6yrs or 53yrs ago. Ruby has weekly heel prick tests and we also have DPNH at home for early detection of high levels. We are very lucky here in Australia as all special food is imported from America and is subsidised by the Australian government.

We have been very happy with Ruby's development to date. She started walking at around 11months and doesn't seem to have lost anything to the kids at day care or mothers group. We have real hope for Ruby's future now. Meeting mums, Uni students, Uni grads and hard working MSUD sufferers has really helped.

We are in the middle of winter here which is very mild compared to Chicago but it is playing havoc with Ruby. She has been basically unwell with some sort of illness now for around 3 months. We battle on and keep a close eye on her levels. We know we will get there, especially now with the network of people we can draw on for advice.

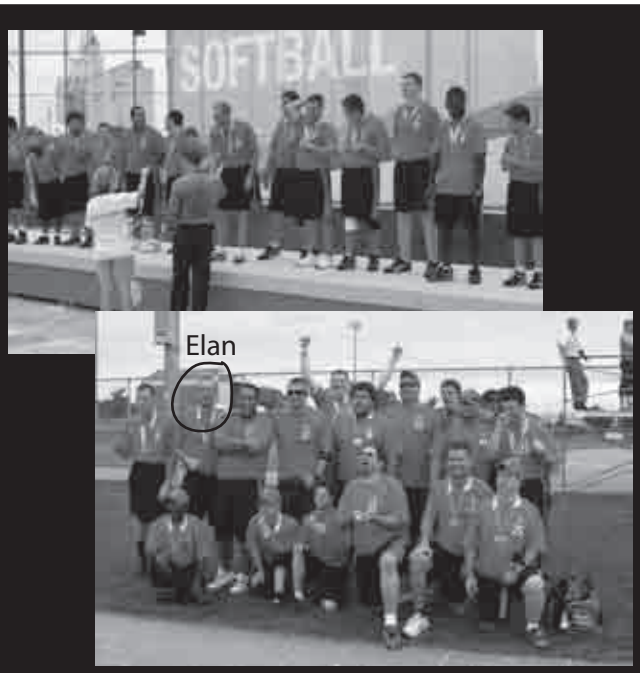
Fiona, Ruby and I would like to thank everyone involved with the MSUD family support group, Joyce, Wayne, Janine and Barbara for every thing they have done and all the support these guys receive to make every thing happen. xxx ■

Genetic Counseling and MSUD

By Randi Zinberg, MS,CGC and
Louise Bier, Graduate Student
Mt. Sinai School of Medicine

The care of individuals with Maple Syrup Urine Disease involves a number of medical specialists. Genetic counselors are members of the team who can play an important role in helping a family adjust to a diagnosis of MSUD.

Genetic counseling is "the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease… [it] integrates interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; education about inheritance, testing, management, prevention, resources and research; and counseling to promote informed choices and adaptation to the risk or condition" (National Society of Genetic Counselors, 2010). Genetic counselors are experts on the genetics of MSUD and other genetic conditions, and are also experienced counselors who are able to provide support to individuals and families affected by a genetic disease. The goal of the genetic counseling process is to provide families with accurate and understandable information, and to help them make decisions that are appropriate for their own unique needs.



Elan Geffen's (transplanted, 2006) softball team, The Wildcards, won a Bronze Medal at the 2010 Special Olympics National Games in Lincoln, Nebraska this summer!

In most cases, the diagnosis of MSUD is unexpected, and parents are overwhelmed as they are presented with an enormous amount of new information. As genetics educators, genetic counselors are able to break down complicated information into more manageable pieces. They can help interpret the information in a way that is meaningful to family members. A typical genetic counseling session can last an hour or more, allowing time for families to ask questions, address concerns, and ensure a thorough understanding of the issues. MSUD is a genetic disease, and an individual's diagnosis has implications for other family members. By taking a detailed family history, a genetic counselor can help identify other family members who may be at risk. A genetic counselor can help families think about how to tell other relatives about the diagnosis and what it might mean for those relatives. If relatives that the family chooses to inform have questions, the genetic counselor can meet with them to address their issues.

There are times when a family with a new diagnosis of a genetic condition is not helped by facts and figures, but instead needs to react emotionally to the diagnosis. It can be a confusing and isolating time. A genetic counselor's role is then to provide supportive counseling to the family, explore the meaning of the diagnosis and, if desired or needed, help arrange other types of support, including private and group support, and other networks to help families adjust and incorporate the diagnosis into their lives.

The role of a genetic counselor can continue beyond the initial diagnosis as the needs of families change. As a child with MSUD gets older, he or she may start asking more questions about MSUD and what it means for them. A genetic counselor can help parents prepare to answer these questions in a way that is appropriate for their child and that encourages further open communication. Parents thinking about having another child may have questions about the risks of recurrence and how to address those risks. Genetic counselors can provide this information and present all the available choices to the parents, such as prenatal diagnosis or pre-implantation genetic diagnosis. Again, the genetic counselor can support a couple as they consider their options and encourage them to reach a decision that is appropriate for their needs. Further, as the body of knowledge about MSUD grows, a genetic counselor can also help families interpret new scientific information and can help them get involved in appropriate research studies, if desired.

In summary, genetic counselors can play an important role in the care of families with genetic conditions through education, counseling and support. To find a genetic counselor in your area, talk with your doctor or visit the National Society of Genetic Counselors website at www.nsgc.org. ■

A Few Words From the Editor

Karen Dolins, EdD, RD, CDN
Newsletter Editor

So much has happened since our last newsletter!
Due to my geographical proximity and my RD status, I was asked to represent our group at the Genetic and Metabolic Dietitians International (GMDI) conference in Baltimore on April 15-17. I arrived with brochures describing our support group, and manned a table in the exhibit hall where I met many dietitians from all over the world. I was amazed and thrilled to hear that 5 were in attendance from Australia, 7 from Venezuela, and 2 from Poland! I'm sure I met dietitians who work with many of you. I know I met the Brubachers'!

When I wasn't in the exhibit hall, I attended sessions. I learned that an initiative is underway to develop nutrition guidelines for all of the Inborn Errors of Metabolism, including MSUD. Dianne M. Frazier, PhD, MPH, RD is spearheading this project, and describes it in her article on page 3.

I also learned practical guidelines for helping children with limited diets have a positive eating experience, and heard psychologist Susan Weisbran, PhD report on her work in the area of neuropsychology and Inborn Errors of Metabolism. It was incredibly reassuring to know that so many capable people are working so hard to help affected individuals live up to their potential. Special thanks to Applied Nutrition Corp. for helping to fund my trip.

More recently, Hannah (classic 16 years), Jerry and I attended Symposium 2010 in Libertyville, Illinois. The symposium never disappoints. We met many new families, shared advice, and were motivated to continue our advocacy efforts. Hannah and I decided it was time to take part in a fundraising effort. Barbara Mudrick has agreed to lend us her expertise, we've rounded up extended family members, and we're planning an event in the Chicago area. We hope to raise money for research efforts as well as practical help for MSUD families in need.

This issue is packed with information on the symposium for those of you who were unable to attend. We also have an article on genetic counseling and two on MSUD and pregnancy, one written from the perspective of a mom and the other by dietitian Sandy van Calcar, who teaches us what she has learned helping individuals with MSUD experience safe pregnancies (see page 14). Glenda Groff continues to provide us with recipes to tempt the appetite, and Joyce Brubacher tells us about her experiences in Costa Rica.

Anyone can write for the MSUD newsletter. We all have a story to tell. Writing expertise isn't required, just your willingness to help others through your words and wisdom. Start today by writing down your thoughts, and I'll help you turn it into an article for the next issue.

In health,
Karen Dolins

Formula Crisis in Costa Rica

by Rebecca Kropf

This is a report on the MSUD formula crisis in Costa Rica which began in June, 2009. We were told there would be no more of the Mead Johnson BCAD formula until September. Some children with MSUD were running out of formula and the families shared the little they had left. Each can provided one more week of hope and well-being.

Because my foster son Dwight had died, I had extra formula on hand. [See the article "Our Visit With MSUD Families in Central America" by Joyce Brubacher in the Fall 2009 issue.] Following the doctor's advice, I calculated the amount of formula I needed until the crisis was over for my foster daughter Karla who also has MSUD. I was able to provide 21 cans of the BCAD to share with other families. This was only a drop in the bucket in comparison to what was needed. During this time, there was only one child hospitalized. This was partly due to reflux, although he was also without MSUD formula.

I told Joyce Brubacher about our formula shortage. She put me in contact with Rick Finkel from Applied Nutrition to see if he would donate some Complex Essential formula. He shipped 32 cans free of charge with the shipping paid. The formula came in my name, and by a miracle from God, I was able to get it out of customs in one day. I was grateful for the help of the sympathetic officials who worked in the various offices, otherwise we would not have been able to get it out of customs for 30 days. At the end of that weary day, with the help of others, I delivered 16 of the cans to Dr. Saborío, our doctor at the clinic in San Jose. The other 16 cans we divided between the families most in need, giving 2 cans for each child with MSUD.

We parents are very grateful to Rick and Applied Nutrition for this lifesaving gift. The children really liked this formula. About 2 weeks later, we once again had formula available, MSUD Maxamaid from Nutricia. Finally, in February of this year, we were able to get BCAD again. Hopefully, the ones responsible for keeping formula in stock will plan ahead in the future because the crisis caused untold anguish and distress for the parents as all of you can imagine! ■

A Day of Fun, Food & Raising $$$

The Clinic For Special Children in Strasburg, Pennsylvania, held it's 20th annual auction on Saturday, September 18th. The total amount raised was $357,000 with 1,730 bidders, which the clinic relies heavily on to continue to provide low cost care to it's patients.

Some Auction Facts & Figures...

The Postage Stamp quilt sold for $3,000. It was the 16th such quilt that was created especially for the annual auction, raising a total of $28,000 for the Clinic. A 17th Postage Stamp quilt is already in the works for next year's auction.

Dr. Strauss, the Clinic's Medical Director, made an unusual request that people attending the auction donate a dime per calorie that they eat at the fundraiser. That request brought in $8,000!

A collection of Dr. Morton's (the Clinic's Director), lecture notes and a signed copy of the PBS documentary about the Clinic was purchased for $7,000 by Dr. Stephen Eck, senior vice president for research at Eli Lilly.

Stuffing Balls

1/2 cup chopped onion
1 cup diced celery
1 cup shredded carrots
1 cup butter
1/2 teaspoon sage
1/2 teaspoon pepper
2 tablespoons parsley flakes
1 teaspoon salt
6 cups crumbled low protein bread
1 1/2 cups mashed potato
1- 1 1/4 cups water (can use water left over from boiling potatoes)

Sauté celery, onion and carrots in butter until tender. Mix remaining ingredients with 1 cup water, then adding in 1 tablespoon increments until the mixture in moist enough to shape into balls. Shape into balls using the bread mixture. Bake at 350 degrees for 20 minutes. These stuffing balls can be shaped and frozen in a single layer on a baking sheet. Once they are frozen, store in a tight container and take out as needed. They can also be flattened and fried for a sandwich. Suggestion: Add 1 tablespoon Old Bay Seasoning to mimic a crab cake.
Makes 18 stuffing balls.

	Protein	Leucine	Calories
per recipe	8.4g	1158mg	5222
per serving	.46mg	65mg	290

Please send recipes to
Food News Editor

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Carrot Loaf

1 cup low protein bread crumbs
1/2 teaspoon celery salt
2 tablespoons coffee creamer
1/3 cup warm water
1 cup carrots, cooked and mashed
1 tablespoon butter
1 tablespoons onion, minced

Sauté onion in butter until tender and combine with carrots, bread crumbs, coffee creamer, water, and salt. Place in greased casserole dish and bake at 350 degrees 25 minutes. Makes 2 servings.

	Protein	Leucine	Calories
per recipe	2.3g	128mg	440
per serving	1.2g	64mg	220

Zucchini Quiche

3 cups grated zucchini
1 cup chopped onion
1 cup Cambrooke Foods MixQuik
3 tablespoons Country Sunrise egg mix
5 tablespoons water
2/3 cups oil
1/2 cup low protein cheese shreds
1 teaspoon parsley
1/4 teaspoon salt

Combine all ingredients and pour into a greased 9 inch pie plate. Bake at 350 degrees for 30-40 minutes.
Makes 8 servings.

	Protein	Leucine	Calories
per recipe	7.8g	432mg	1976
per serving	0.97g	54mg	247

Deep Dish Taco Supper

1/2 cup Cambrooke Foods MixQuik
1/3 cup water
Mix well and pour into a greased single serving casserole dish
1/4 cup PKU Perspectives mushroom burger mix
1 teaspoon oil
2 Tablespoon water
Oil
2 Tablespoons diced tomatoes
1 tablespoon diced green pepper
1/4 cup mayonnaise
2 tablespoons shredded low pro cheddar cheese
1 tablespoon minced onion
3/4 cup Shredded lettuce
1/4 cup French dressing

Mix burger mix, 1 teaspoon oil and water together. Heat frying pan and fry in additional oil cut with spatula until browned. Place on top on baking mix. Sprinkle with peppers and tomatoes. Mix mayonnaise, cheese and onions and carefully spread on top. Bake at 350 degrees for 20-25 minutes. Remove from oven, top with lettuce and French dressing, and serve.
Makes 2 large servings.

	Protein	Leucine	Calories
per recipe	2g	220	1100
per serving	1g	110	550

I wanted to follow up with all the people I met at the MSUD Symposium in Chicago. I appreciated the opportunity to come and speak with you. Again I must admit that I was completely unaware that this disease even existed, and the education process on it has been very unique.

All the families afflicted with this disease face issues that cannot be imagined by the laymen public. The daily struggles that you face are beyond comprehension by anyone who has no real knowledge of it such as myself. You have watch loved ones fight on a regular basis with diets, medical procedures, and the daily norms of life, which can never be viewed as normal by the general public, because most people take those things in everyday stride. You are all truly remarkable individuals

My hope is that in the sixty minutes I was on stage, you were able to take something away from my presentation that will help you in your daily life, and aid you in the struggle that is this disease. Please remember what I said to you. This disease is ONLY an affliction. IT IS NOT WHO YOU ARE! Do not let it consume you or your way of life. Do not measure your self worth by it. Do whatever you need to do to find yourself, and rise above the disease. Hold on to God's unchanging hand and pray, and never forget, always remember to:

"DREAM AS IF YOU ARE GONNA LIVE FOREVER, AND LIVE AS IF YOU ARE GOING TO DIE TODAY".

Good Luck to all of you and God Bless You!
John Parker, Motivational Speaker

Brendan Lee, M.D., Ph.D., Professor, Department of Molecular and Human Genetics, Baylor College of Medicine and Investigator, Howard Hughes Medical Institute.

Research Update: Treatment of MSUD with Phenylbutyrate

As reported in our Winter 2010 issue, Dr. Brendan Lee has been applying lessons learned treating urea cycle patients to those with MSUD.

It has been observed that levels of the branched-chain amino acids and their keto acids are lowered in patients treated with phenylbutyrate. This treatment was tested in 5 patients with mild presentations of MSUD. Results were variable, but some effect was observed. Given this encouraging news, Dr. Lee hopes to conduct a clinical trial in which patients with classic and intermittent MSUD will be treated with either phenylbutyrate or placebo in a crossover design (each subject will receive each treatment). This will be a 4 week study with a 2 week washout period (neither drug nor placebo) in between the trials.

Dr. Lee notes that phenylbutyrate is already FDA approved. If it is found to be effective in increasing tolerance to protein and/or preventing or mediating metabolic decompensations, approval for use would involve a label application which takes about 90 days. ■

2010 MSUD Symposium

Symposium attendees included this group from Israel, Brazil, Peru, Costa Rica, Hawaii, Honduras, Australia and 3 States on the U.S. mainland, some of whom were grateful to receive support from our group allowing them to attend.

LEANNA PETERS' SECOND PREGNANCY

Written by Mary Ann Peters, Leanna's mother

Five years after a difficult first pregnancy, my daughter became pregnant with her second child. This pregnancy came as a surprise to me as she knew the risks associated with pregnancy.

For the first 4 months, she went to the local OB/GYN office and was then referred to Yale Maternal Fetal Medicine in New Haven, CT where they manage high-risk pregnancies. As with her first pregnancy, Leanna experienced morning sickness but this time it was for a much shorter period. Overall, she felt pretty good until the last month, and then she became more uncomfortable with every passing day.

Unfortunately I cannot give you information regarding her leucine levels because she did not have blood work taken very often. Leanne has never been compliant with her diet. She continued eating foods that she was not allowed to eat and did not drink her formula every day. Not surprisingly, when she finally started sending in blood work, her amino levels were always elevated.

As she got closer to her due date, the doctors at Yale decided to schedule her for a C-section to avoid the stress of labor. Her official due date was April 28th but her C-section was scheduled for April 21st. On the morning of April 21st, we arrived at the hospital and they prepped her for the c-section. I was very nervous watching her being wheeled into the operating room because I kept remembering the first time we were in this situation and all the drugs she was given to make the labor progress for a vaginal delivery before they realized a C-section was needed. I kept pacing the floor and looking at the clock hoping she would be out soon. After 2 hours, she was wheeled out to the recovery room fully alert and holding her new baby boy. She commented that this time she was able to actually hear the baby cry when he was born unlike her first time. After several hours in recovery, she went up to her room and remained in the hospital for 3 days.

During her hospital stay, I brought MSUD II to the hospital so she could continue drinking it, and the nutritionist tried limiting her protein intake to 8 grams a day. Leanna was always eating more protein than the doctor recommended and her leucine levels were always elevated.

Leanna and baby were discharged on the 4th day and she continued to feel good. When she returned home, she decided to eat low protein foods and continue drinking her formula because she realized it made her feel better. Unfortunately this only lasted a few days before she went back to eating any foods she desired and not drinking her formula every day.

Fortunately, both of Leanna's children are healthy.

I have discussed liver transplant with her. Leanna says she is afraid she will die during the surgery. She also has a problem swallowing pills and gags which could be a serious problem with taking anti-rejection medication. Every day is a challenge with Leanna and I hope some day soon she will finally realize she must stay on her diet and drink her formula to feel her best. ■

Jordan at the Eifel Tower

The Bulchers Visit France

By Sandy Bulcher

Last summer, my husband Dave and I entertained the idea of taking Jordan, our 20 year old son with MSUD, on a European vacation. Our older son, Tyler, had had the opportunity to travel abroad during college and we felt that it would be great if Jordan had the opportunity also.

I admit that the logistics of planning a vacation outside the US was somewhat frightening initially. However, we have always believed that, as much as possible, MSUD should not limit Jordan's life experiences. With that being said, his health and safety is always our first priority.

(*VisitingFrance* cont. on page 13)

(*VisitingFrance* from page 12)

This past winter, we shared our plans with Jordan and, of course, he was excited about traveling to Europe. Jordan had studied French language, history, and culture for several years, so a trip to France seemed logical. After researching various tours, we chose an 8 day trip to Paris and the countryside of France.

In advance of our trip, I contacted Jordan's metabolic specialist to obtain contact information for a metabolic doctor in Paris. In addition, I obtained a letter from his MD that briefly described MSUD and what supplies we would be traveling with on the plane, in hopes of avoiding delays through security. I also gathered MSUD literature including how to treat a metabolic episode. Our travel agent contacted the tour company and informed them of Jordan's medical condition, including the need for low protein foods and refrigerators at our hotels. And finally, Jordan had his branched amino acids drawn to ensure that he was in good metabolic control before our departure.

At the airport, we did not risk checking the bag that contained the MSUD powder and other supplies for making formula for fear that it could get lost and not arrive in Paris. Instead, Jordan carried that bag with him on the plane. He made enough formula for two days and also carried that in a small cooler on the plane, along with some protein free candy. Security at the Columbus, Ohio airport looked through his bags and waved litmus paper over his formula to ensure that it was safe to take on board. Overall, the process of getting through airport security went smoother than I had anticipated.

Our week in France went great with no medical issues or problems. I was concerned that lack of sleep and change in time zones may affect Jordan's leucine level, but fortunately that was not the case. We enjoyed the beautiful city of Paris and many historical attractions outside of Paris.
Our tour director was especially helpful. She ensured that Jordan had appropriate food. Others on the tour group were jealous of the wonderful salads and vegetables that he was served.

Even though we were somewhat apprehensive about taking Jordan on a vacation outside the US, I am glad that we decided to move forward with our plans and allow Jordan the opportunity to travel to France. Hopefully, we will be able to take another trip abroad in the future. ■

Advocating for Better Lives

Kelly McDonald is the Advocacy Chair of the National PKU Alliance. The following article is based on her presentation:

The mission of the National PKU Alliance is to improve the lives of individuals and families with PKU through research, support, education and advocacy, while ultimately seeking a cure. Kelly spoke to our group about the need for concerned people to advocate for individuals with metabolic diseases, as it is only through advocacy that laws are changed.

The Genetic Information Nondiscrimination Act was signed into law in May 2008. It is the first legislative expansion of the Equal Employment Opportunity Commission since the passage of the Americans with Disabilities Act (ADA) in 1990. This law prohibits discrimination by health insurers and employers based on an individuals' genetic information, including genetic testing and family medical history. It affirms that all people have the right to be judged according to their ability to do a job, not on stereotypical assumptions.

The Patient Protection and Affordable Care Act (PPACA) was signed into law on March 23, 2010.
Children with pre-existing conditions can no longer be denied health insurance coverage. By July 2010, all Americans who are uninsurable because of a pre-existing condition will have access to insurance through a state administered temporary high-risk pool, until the private health insurance exchanges are in place in 2014. Insurance companies will be banned from placing lifetime or annual limits on coverage or canceling coverage due to illness, and it allows parents to extend coverage for their children up to their 26th birthday.

The Medical Foods Equity Act (S.2766 and HR.4926) would mandate that ALL insurance companies must provide coverage for medical foods (including formula, foods modified to be low in protein), and equipment, over the course of a lifetime. The bill currently has 23 sponsors. Your help is needed to make sure this important legislation is passed.

Grassroots advocacy is essential to keep us moving forward. We are all urged to call and email our elected representatives, write letters-to-the-editor, and visit members of Congress. For more information on how you and everyone you know can help, go to: www.npkua.org ■

MSUD and Pregnancy:
The Importance of Dietary Treatment

Sandy van Calcar PhD, RD
Senior Metabolic Dietitian
Biochemical Genetics Program, Waisman Center
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The Biochemical Genetics Program at the University of Wisconsin in Madison has followed 6 pregnancies in women with MSUD (3 mothers – 2 with classical MSUD and one with variant MSUD – each mother has had 2 children). We are currently following another woman who is in her second trimester. There have been reports of a couple other pregnancies in those with MSUD, but experience is still limited and the metabolic community needs to continue to learn more about pregnancies in this disorder (1-3).

At this time, we do not have enough information to determine if leucine or the other branch chain amino acids are teratogens when levels are higher than normal. Teratogens are compounds that, when ingested by a pregnant woman, can have adverse effects on her developing fetus. Commonly known teratogens include alcohol and various drugs that can have devastating effects on an infant's outcome. In metabolics, we know most about the teratogenic effects of phenylalanine (phe), the amino acid that cannot be metabolized in PKU. Pregnancies in PKU have been well studied and it is clear that elevated phe levels during pregnancy can cause microcephaly (small head size), developmental delay and congenital defects.

We do not know if leucine is a teratogen. In our pregnancies and the few that have been reported, infant outcome appears to be good. BUT, our women were on diet before pregnancy, and continued diet treatment throughout pregnancy, and maintained normal blood leucine levels. So we really can't say if the infant outcome would be different if this had not been the case.

Women with MSUD who are pregnant continue to be at risk for metabolic episodes associated with illness or poor intake. Pregnancy does not reduce this risk. The first trimester has been especially difficult for some of our women because of morning sickness. The nausea and vomiting associated with morning sickness can make it difficult for a woman to take sufficient calories and protein from formula. This can lead to metabolic decompensation, just as it can with women with MSUD who are not pregnant.

Another time that we have found that women are especially vulnerable to metabolic decompensation is during delivery and the post delivery period. After delivery, a woman begins to "break-down" protein stores as the body changes from pregnancy metabolism to post-pregnancy metabolism. We have found that it is imperative that a woman follow her diet and continue to be monitored for a minimum of 2 weeks after delivery to prevent a metabolic episode. The importance of continuing diet after delivery was especially evident in a research report from Japan published in 2003. This woman was not very compliant with diet treatment during pregnancy, but stopped treatment after delivery. This woman died 51 days after delivery (3). This clearly shows the importance of diet management during and after a pregnancy in MSUD.

There are women with mild forms of MSUD that who may not require strict diet treatment as an adult and have few, if any, metabolic episodes. BUT, pregnancy is a big stress on metabolism as is the post-pregnancy period. So, even if a woman does not have problems associated with MSUD when she is not pregnant, this does not guarantee that she would not have problems during a pregnancy. Any woman with MSUD, no matter how mild it may be, should be followed by a metabolic clinic before pregnancy to assure good metabolic control and continue to be followed during and after her pregnancy. All of our women have been referred to a high-risk obstetrics clinic for more extensive monitoring of the mother and her developing fetus. ■

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My Child Doesn't Have PKU or an "Allied Health Disorder" – So Why Do I Keep Hearing About Both?

By Christina Sciarrotta – President, PKU Organization of Illinois

When my son Alex was diagnosed with PKU nine days after he was born in August of 2003, I did what all parents whose infants have been diagnosed with a rare disorder do: I set about to educate myself. One of the first things I discovered was the large support network for PKU in Illinois – including a structured not-for-profit support organization.
One year later, I attended a conference in Michigan in which the topics and audience were not solely related to PKU, but other similar, yet rarer disorders. That was my first exposure to some of the stories and details around other metabolic disorders – or what we've come to call Allied Health Disorders.

I joined the PKU Organization of Illinois as a Board Member in 2005 – the same year our by-laws were revised to expand our membership guidelines to include not only individuals with PKU and their families, but now including individuals and their family members with the following inborn errors of metabolism: Urea Cycle Disorder, Maple Syrup Urine Disease, Homocystinuria, Tyrosinemia, Glutaric Acidemia – Type 1, Methylmalonic Acidemia, Propionic Acidemia and Organic Acidemia. PKU is the most common of this group.

As part of this community, you may have heard about the PKU Organization of Illinois or our events in one form or another and been confused about what an "Allied Health Disorder" is and how it applies to you. When the PKU Organization expanded its focus to the eight other inborn errors of metabolism, the term we used to refer to those eight was "Allied Health Disorder." We use "Allied Health Disorder" in many of our communication forums and event marketing to refer to any of the eight inborn errors of metabolism listed above.
The larger question of course is why did we expand our focus and how does it benefit you? To answer to this is simple….and complex.

The reason why our organization expanded its focus is simple – as the most common inborn error of metabolism, we have a fairly large community. One large enough to build a successful and effective local support organization (many other states have similar non-profit PKU support organizations). While MSUD (and some of the other conditions) have national support networks, it doesn't have a local support group to take advantage of closer resources on a more frequent basis. And both Illinois genetics clinics we work with recognized similarities in support needs across PKU and the other rarer conditions. With the support structure already established, it made sense to expand our organization and ultimately our network of resources, to be able to assist families dealing with similar challenges.

So how does this benefit you? That answer is more complex. While we've been working hard to reach out to the "Allied Health Disorder" Community to let you know who we are and what we do, we haven't always answered the question of how specifically we can support you – or highlighted which of our events or resources apply to you and which may not. Because - while we have similarities, there are also differences. We've been working hard this year to tailor our communications to the Allied Disorder Community in a way that answers these questions, so ultimately we can support you not just in our by-laws, but in a tangible way.

The main thing you should know about the PKU Organization of Illinois is that our charter is two fold – to raise funds for research and to provide community support. Our standard yearly calendar of events in Illinois includes a New Parent Coffee, Low Protein Cooking Classes, Weekend Camp and an Annual Meeting which features speakers and breakout sessions on metabolic disorder related topics. All of these can be great networking and education resources for you. We've also recently aligned with the newly formed National PKU Alliance. NPKUA is fighting hard for a Food Equity Act in Washington, which includes 29 inborn errors of metabolism. Our help supporting NPKUA with their vision will ultimately benefit the entire inborn error of metabolism community.

Please visit our website at www.pkuil.org for more information on us, our vision and events. We'll continue to provide specific information on how we can support you and we look forward to seeing many of you at our Annual Meeting in November. ■

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