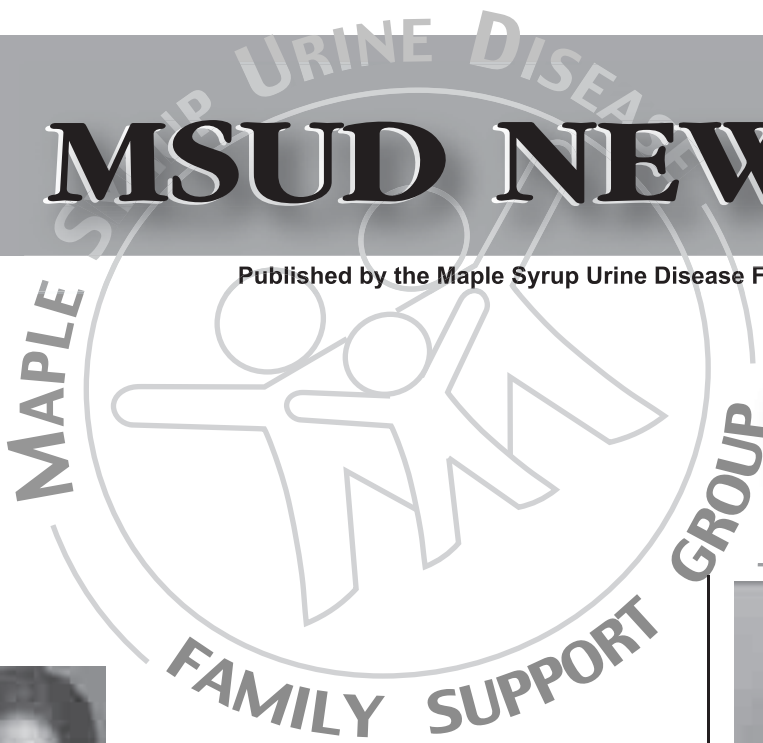


MSUD NEWSLETTER

Published by the Maple Syrup Urine Disease Family Support Group • Volume 25, Number 1 • Spring • 2008



MSUD Symposium 2008
June 26-28, 2008
Columbus Embassy Suites, Columbus, Ohio
Register now! See page 3.



Welcome to the Newsletter

Karen Dolins, EdD, RD, CDN
Mom to Hannah, Age 13

I hope you enjoy this issue of the newsletter. It is meant to provide updates in research and treatment, stories from member families, and practical information such as diet tips and new food products.

A new newsletter has its genesis as soon as the previous one is published. Requests are made to professionals and families alike for contributions. As everyone is busy, it takes quite awhile to gather enough information to publish. But the result makes it all worthwhile.

We are a family. I still remember the first newsletter I read after my daughter Hannah was diagnosed in 1994. It allowed me to feel connected to a group of people, to know that I was not alone, and that others would help me.

This issue features moving stories from members. The heartwarming story of the Burkholder family, with 12 children, 6 with MSUD puts into perspective the issues those

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Page 6,7 PLIGHT OF MSUD PATIENTS IN THE PHILIPPINES

The information contained herein does not necessarily represent the opinions of the MSUD Board, Medical or Nutritional Advisors, or all of our members. Before applying any of the information contained in this newsletter, you must consult a MSUD specialist.

TOPIC:
Medical Research



Oxidative Stress in Maple Syrup Urine Disease

Peter J. Mc Guire, MS, MBCh
George A. Diaz, MD, PhD

It's a part of life. We breathe in oxygen and breathe out carbon dioxide. The oxygen is used to get energy from the breakdown of sugar or fat molecules, and the carbon dioxide removes waste. During this life-sustaining process, a small amount of oxygen is converted into free radicals, for example reactive oxygen species (ROS). Fortunately, our body has an antioxidant defense system which detoxifies free radicals and prevents them from causing damage. If the defense system can't keep up with demand or if there is an overproduction of reactive oxygen species, the result is "oxidative stress," a term which refers to the accumulation of potentially toxic forms of oxygen and other atoms beyond the body's capacity to detoxify them.

During periods of oxidative stress, the toxins can damage various cell components, including proteins, fats in cell membranes, and DNA by reacting with and altering their chemical composition. When this happens, cellular functions can become progressively impaired. When the impairment of cellular function is widespread, the effects can be detected at the level of tissues and organs. A theory has been proposed (the free radical theory of aging) that oxidative stress leads to

(Oxidative Stress cont. on page 2)



(Oxidative Stress *cont. from page 1*)

the gradual accumulation of cellular damage and subsequent tissue degeneration associated with aging. In support of this hypothesis, oxidative stress has been demonstrated in many different disease processes including diabetes, cancer, heart disease, Alzheimer's disease.

A number of nutrients aid in the antioxidant process, including vitamins C, E, and beta carotene. Supplementation of these nutrients has become relatively common, though the benefit of these supplements is not well established and there is some evidence that at high levels they actually become pro-oxidants, creating more free radicals instead of detoxifying them.

In studies of several different inborn errors of metabolism, evidence of oxidative stress (increased levels of ROS combined with a decrease in antioxidant defenses) has been reported. Laboratory studies of clinical samples from patients with MSUD, as well as animal models of MSUD, have suggested that increased levels of oxidative stress occur in this disease. The cause of the oxidative stress is still being debated. However, it has recently been suggested that in MSUD, as well as in other inborn errors of metabolism, the accumulation of compounds resulting from the inherited enzyme deficiency "poisons" the normal processes occurring in the mitochondria, resulting in the increased production of ROS which overwhelms the antioxidant defense system. Indeed, decreased levels of blood antioxidants have been described in patients with MSUD. Vitamin E (also known as

tocopherol) was found to prevent the "mitochondrial sickness" induced by treating rat brain slices with high levels of branched-chain amino acids (valine, isoleucine, leucine) seen in an animal model.

Despite these interesting preliminary studies, the role of oxidative stress in MSUD has not been established definitively. While the role of oxidative stress in common diseases has been an area of intense research activity, investigation of the role of oxidative stress in inborn errors of metabolism has not been as extensive. It has been postulated that some of the neurologic features seen in MSUD may be due to oxidative stress. However, these speculations must be treated with caution as they require further validation. Moreover, the benefit of antioxidants in inborn errors of metabolism is also unproven. Conflicting studies in the literature on the effect of antioxidants in heart attack and stroke suggest that the role of antioxidants in human disease needs further evaluation. Due to the potential for pro-oxidant effects of certain antioxidants, their use via supplementation in MSUD cannot be recommended at this time. A diet containing fruits and vegetables serves as a source of natural antioxidants.

The results of recent work supporting a connection between branched-chain amino acid metabolism and oxidative stress suggest that further research may provide new insights into the pathways involved in the MSUD disease process. It also emphasizes the importance of including ample fruits and vegetables in the diet (see accompanying article on "Getting More Essential Nutrients into the MSUD Diet" by Alex Larkin, RD). ■

References

- Barschak, A. G., A. Sitta, et al.** (2006). "Evidence that oxidative stress is increased in plasma from patients with maple syrup urine disease." *Metab Brain Dis* 21(4): 279-86.
- Bridi, R., J. Araldi, et al.** (2003). "Induction of oxidative stress in rat brain by the metabolites accumulating in maple syrup urine disease." *Int J Dev Neurosci* 21(6): 327-32.
- Bridi, R., C. A. Braun, et al.** (2005). "Alpha-keto acids accumulating in maple syrup urine disease stimulate lipid peroxidation and reduce antioxidant defences in cerebral cortex from young rats." *Metab Brain Dis* 20(2): 155-67.
- Bridi, R., A. Latini, et al.** (2005). "Evaluation of the mechanisms involved in leucine-induced oxidative damage in cerebral cortex of young rats." *Free Radic Res* 39(1): 71-9.
- Ribeiro CA, Sgaravatti AM, Rosa RB, Schuck PF, Grando V, Schmidt AL, Ferreira GC, Perry ML, Dutra-Filho CS, Wajner M.** Inhibition of brain energy metabolism by the branched-chain amino acids accumulating in maple syrup urine disease. *Neurochem Res*. 2008 Jan;33(1):114-24.
- Pérez de Obanos MP, López-Zabalza MJ, Arriazu E, Modol T, Prieto J, Herraiz MT, Iraburu MJ.** Reactive oxygen species (ROS) mediate the effects of leucine on translation regulation and type I collagen production in hepatic stellate cells. *Biochim Biophys Acta*. 2007 Nov;1773(11):1681-8.
- Funchal C, Latini A, Jacques-Silva MC, Dos Santos AQ, Buzin L, Gottfried C, Wajner M, Pessoa-Pureur R.** Morphological alterations and induction of oxidative stress in glial cells caused by the branched-chain alpha-keto acids accumulating in maple syrup urine disease. *Neurochem Int*. 2006 Dec;49(7):640-50.



DON'T BE LEFT OUT!

Become a member of the MSUD egroup. The only requirement is that you are a member of the MSUD Family Support Group. To join the MSUD eGroup, send an email to the moderator, Emily Talley at emilytalley@mindspring.com requesting to join the egroup.

To become a member of the MSUD Transplant eGroup go to the link <http://health.groups.yahoo.com/group/msudtransplant/> and search for msud and liver transplant. You can also email Oula Haddad at jhaddad1@yahoo.com requesting to join the group.

MSUD SYMPOSIUM JUNE 26 - 28, 2008

Plans are underway for MSUD Symposium 2008. I can't wait to meet new families, renew old friendships, and learn more about MSUD. Like past conferences, we're expecting a large number of attendees.

Hope to see you there!
Sandy Bulcher

WHERE Columbus Embassy Suites

2700 Corporate Exchange Drive, Columbus, Ohio 43231
614-890-8600 • 1-800-Embassy (362-2779)

Each suite has a private bedroom with 1 king or 2 double beds and a separate living area with a sofa bed, dining table, refrigerator, and microwave. The hotel has an indoor/outdoor pool, whirlpool, sauna, and fitness center.

NOTE This is not the same Embassy Suites where Symposium 2006 was held (this hotel is located closer to the airport and provides free shuttle service to and from the airport)

WHO MSUD families, friends and professionals

COST Registration for the conference is free to all attendees

Hotel rate: \$114 per night, includes free parking, buffet breakfasts and evening snacks. Lunches on Friday and Saturday are provided by the MSUD Family Support Group. Low protein food will be provided. Dinners are on your own. An on-site restaurant serves mainly American cuisine.

TRANSPORTATION The hotel is located on the north side of Columbus with easy access to the outer belt (I-270) and I-71. Directions from I-270: Take Cleveland Ave South to Community Park Drive. Turn left on Community Park Drive and go one block to Corporate Exchange Drive. Turn left onto Corporate Exchange Drive and the hotel is on your left. Port Columbus, the nearest airport (airport code CMH) is 15-20 minutes from the hotel

Complimentary shuttle service is provided by the hotel. This service can be used to and from the airport, local restaurants, and shopping.

Those needing travel assistance, see bottom of page 4 of this newsletter.

SCHEDULE Thursday June 26th: Registration/Reception 7 - 9 pm

Friday June 27th: Meeting times 9 am - 5 pm

Saturday June 28th: Meeting times 9am - 5 pm

TOPICS TO BE ADDRESSED Medical and Dietary Management, Adult Metabolics, Liver Transplantation, Research, Sibling Issues, and more!

IMPORTANT It has become increasingly difficult to provide adequate childcare, due to the large number of attendees. Because of this, we need to ask parents to provide for their own children. Please take this into account as you plan your trip. Consider bringing your own babysitter (grandparent, teenager, friend) or taking turns with your spouse.

If you have any questions, feel free to email or call at dbulcher@aol.com or 740.548.4475

To attend the symposium complete and return the registration brochure included in this mailing.

(Editor's Message *cont. from page 1)*

of us face with 1 MSUD child. The magical story of Elan Geffen's trip to Israel, and the humorous face that Jordan Bulcher puts on life with his disease will touch us all.

We have a peek into new research being conducted at Mt. Sinai Medical Center in New York, which meshes with a dietitian's perspective on the importance of including fruits, vegetables, and a source of omega 3 fatty acids in the MSUD diet.

Meanwhile, please keep in mind that "it takes a village." You may have noticed that our website has not been kept up to date. You probably don't realize that Sandy Bulcher practically single-handedly puts on our semi-annual symposium, a feat that is usually accomplished by a dedicated committee of people. You will read the touching story of how several of our members have given of themselves, my daughter Hannah and Kay Larson in particular, to provide formula for families in the Philippines who otherwise would have gone without.

As you read through this issue of the newsletter, please don't be a bystander. Be a participant. We need help with the symposium, with our website, and with a myriad of other tasks that are necessary to keep us a vibrant organization.

As winter turns to Spring, our children are growing up, mine included. Come December, we'll be a family of teenagers as my twins turn 13. With growth comes independence. Our daughter Hannah (Classic MSUD – almost 14) is preparing to go away to camp for a full summer! She will be nearby so she can get to the hospital if necessary and my husband Jerry and I will be delivering her formula weekly and monitoring food logs, but it's definitely a milestone for us. Since her siblings began going away to summer camp, she has longed to do the same.

Tell us about your families. Help us with our organizational needs. We inspire each other to reach beyond our perceived boundaries. ■

The following college entrance essay was humorously written in response to the question: If you could travel in time, where would you go and why?

In Search of Sweets

By Jordan Bulcher



The future: an object of society's imagination, hopes, and dreams. Countless science-fiction films have told what we as humans wish the future to be; however, for me it is not about flying space crafts or holograms, but merely a quest for luscious treats. If I had the ability to travel through time, I would venture to the future in search of a cure for Maple Syrup Urine Disease, a rare metabolic disorder, which ravages my life, and strangles my ability to eat those luscious treats.

Over the years, I have watched a myriad of people gulp down shakes, plunge into Dairy Queen Blizzards, and gorge themselves on chocolate. With limited enzyme activity, I have not had this opportunity and it preys on my mind daily. This, of course, is why I would travel to the future; to run rampant through Steak 'n' Shake; to eat, sleep, and breathe Dairy Queen; and most importantly, indulge myself in the every-tasty treats from the Hershey's Chocolate Factory.

After relishing in the aforementioned goodies, my life would be complete and I would feel as though I am on top of Mount Everest.

Until time travel is perfected, I will just have to wait outside of the ice cream shops, nibbling on broccoli, and attempting to swallow a handful of peas.

Travel Assistance to Attend Symposium 2008

Another donation from the United Services Foundation allows the support group to help with travel expenses for families and individuals with MSUD from the United States or any other country who do not have other financial resources available. Those who have never attended a MSUD Symposium will have priority for assistance although anyone may submit a request. If traveling from another country, the process to obtain a visa and make flight arrangements must begin as soon as possible. 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 wishes to attend. Contact Wayne Brubacher NOW at 574-862-2992 or e-mail: wjbrubacher@afo.net.

Matthew Chapman Age 10

By Amy Chapman (Mom)

When my husband Bryan and I had Matthew on February 14th, 1998, he was a perfect baby. Eventually my sister noticed that there was something wrong. He wouldn't move his head for noise, eat, or let me breastfeed. When he lay on his back he would let out a long cry. He slept in his car seat for awhile with his eyes fixed. I was getting very scared and decided to take him to the E.R. He stayed in the local hospital in Taylor County for 3 days but they didn't know what to do for him. Finally, his pediatrician decided to send him to Kosair Hospital in Louisville, Kentucky where he stayed in the ICU for about 2 weeks. By the time he was diagnosed with MSUD, his levels were in the 2,000s, he was in a coma and his brain was swelling. He was finally treated and after about a month we were able to go home.

Matthew now is in 3rd grade and just turned 10. He is doing well and has not been in the hospital for 3 years. He is wearing glasses so he can see better. My husband and I were scared to have another child, but 2 years ago I gave birth to Hanna, who does not have MSUD. Matthew loves her and both children are healthy.

Since Matthew is older it is easier for us to do things, but it is hard to eat in restaurants because of the way they cook. At home he gets all the formula and foods he needs. He is learning each day how to take care of himself but he still doesn't understand why Hanna can have what we have and he can't. He always says "I wish I could have that."

It is hard sometimes. We learn more and more each day. I have to thank his doctors and nutritionist for all their help. The staff and kids at school all know about Matthew, and they call me to ask if it is OK for him to have cup cakes or other things. I have learned to eat what Matthew eats and I try to encourage him to eat his food. I always tell him that God made him special and we love him and want him to take care of himself.

And this is Matthew's Life..... ■



Ross Becomes Abbott Nutrition

Ross Products Division is undergoing a name change. Our new name, Abbott Nutrition, clearly defines who we are and the work we do. As our new name now indicates – nutrition is our business. Although the name on the door has changed, we will continue to provide you with breakthrough products and the very best service.

Abbott Nutrition MSUD Food List Now Available

Abbott Nutrition is offering a new Food List for Maple Syrup Urine Disease (MSUD) booklet!! This valuable resource provides a comprehensive list of baby foods and table foods to help you manage your child's MSUD diet. The 85-page booklet also contains recipes, resources and other helpful information. The Food List for MSUD booklet is a supplement to Abbott's "A Guide for the Family of the Child with MSUD". For more information on how to obtain these booklets, please call your healthcare provider.

Editor's Note

This pocket-sized guide is printed on laminated paper allowing it to sit on your kitchen counter without worries of getting dirty from food. It includes basic information on the BCAA-restricted diet for MSUD. Allowed foods are categorized according to food groups, and nutritional information is based on both gram weight and household measures.

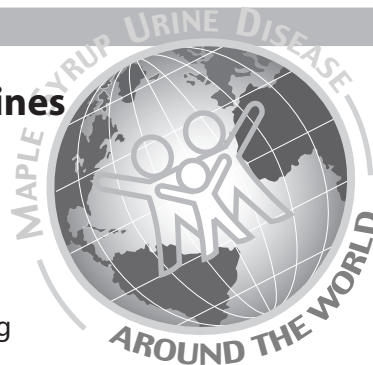
The Quest to Survive: the Plight of MSUD patients in the Philippines

By Dorie Marco

The first MSUD patient in the Philippines was diagnosed in 1983 at the Philippine Children's Medical Center, a government specialty tertiary hospital for children, by Dr. Carmencita D. Padilla, the only clinical geneticist at that time. Over the next twenty years, approximately 60-70 patients have been diagnosed with MSUD. Only 21 of these have survived, now ranging in age from 0-16 years old.

In the Philippines, medical management of an MSUD child is indeed a big challenge due to the following: lack of screening services, unavailability of the medically indicated special powder [editor's note: medical food or formula] and amino acid supplements and other technical resources. Earlier, whole blood was sent to Australia to confirm the diagnosis. When my first child was diagnosed in 1993, blood was still sent to Australia for leucine monitoring. In the mid to late nineties, the Institutes of Human Genetics, National Institutes of Health, UP began offering laboratory services for leucine and plasma amino acid monitoring in Manila.

In a third world country like the Philippines, the majority of families with MSUD don't have the money needed to meet the medical needs of their children, including the special diet powder and special food. Many parents are minimum wage earners, and others are unemployed. Commercially prepared low protein foods such as biscuits and the like can only be availed through the courtesy of relatives residing in the US or Australia, as the high cost of these imported items and the added cost of freight and taxes make them too expensive for most. For example, a can of MSUD powder (400 gms) cost P2,500- 3,000 or \$55-65 American dollars. At present, we are not receiving any subsidy from our government.



Dr. Carmencita Padilla, feeling the burden of the families with MSUD Children, tried to find ways of helping parents by networking with local formula companies and the Department of Health. Through her efforts, Mead Johnson Philippines committed to facilitate the donation of MSUD diet powder to affected children from their mother company based in the US. For the past thirteen years, Mead Johnson Philippines was so kind enough to supply 6 cans of formula monthly to each child with MSUD. While not sufficient to completely meet the medical needs of our children, it was a big help. In order to make the formula last for a month, parents find local alternatives such as using rice soup or "Am". As our children are growing, there is a need for other sources of food aside from formula. What is worse is that we do not have commercially low protein solid food available in the local market. Parents make use of indigenous food just to have something to feed their children.

In 2006, two families with MSUD children were given the opportunity to attend the MSUD symposium held in Ohio through the financial assistance of the MSUD Family Support Group. The participation of a Philippine delegate to that symposium inspired the 2 families to organize the MSUD Philippine Support group. The group was formally organized in August 2006. One of its activities was to help patients undergo plasma amino acid testing. While this essential test is locally available, it is not affordable to most of the patients.

(Philippines cont. on page 7)

(Philippines cont. from page 6)

In April 2007, unexpected news from Mead Johnson Philippines caught parents by surprise and caused us dismay. We were informed that there will be no regular supply of MSUD diet powder for 4 months for two reasons: first, Mead Johnson US was requiring more documents to support the donation; second, the new Philippine Milk code stipulates stricter rules for any form of donations made by any formula companies in the locality. Parents were so helpless and worried for their kids. The local support group sought help from MSUD Family Support Group members in the United States through e-group e-mails.

Our pleas were soon granted. Applied Nutrition Corp. donated 15 cases or 60 cans of Complex MSUD Vanilla, and a parent in the US contributed the shipping fees. The formula was divided equally among our MSUD children. After a couple of months, another 100 cases or 400 cans of SHS MSUD MAXAMAID artificially orange flavored powder was donated by SHS North America through the MSUD Family Support Group in America. The generosity of the two entities prolonged and saved the lives of our children. This lightened the burden and worries of all parents. Volunteers from the MSUD Family Support Group in the States exerted extra effort, patience and time in processing all the necessary requisites in sending the donated formula. The group also paid the substantial freight expenses. Without the assistance of the MSUD Family Support Group in America, most of our affected kids would probably be in metabolic crisis or might be dead.

Eleven months has passed and Mead Johnson Philippines is still in the process of obtaining approval from the Department of Health for the importation of their donations. We are all praying hard that soon this problem will be solved. We are very much thankful for all the kindness that our fellow parents in the MSUD Family Support Group have extended to us. From all of us here in the Philippines, our deepest gratitude to our fellow parents!!! ■

Editor's note: Between the time this article was written and the publication of this newsletter, another shipment of over 63 cases (257 cans) of SHS MSUD Maxamaid has been shipped and distributed to the Philippine families.

My Trip to Israel By Elan Geffen

Age 23, transplanted 1/06

On January 2, 2008, at 6 am, I boarded an Israil plane with 19 other young adults, many of whom I had never met before. We were off to Israel for a 12 day adventure, as part of the Taglit Birthright Israel program- a free trip for Jewish males and females, ages 18 - 26, funded by the Israeli government, the Jewish Federation and some private donors. We had a smaller group than most other Birthright groups, and a few more staff members, but other than that, it was just like any other Birthright trip- busy from morning til night with sightseeing, hiking, tasting new foods, meeting many people, learning about Israel and about each other. Our tour guide, Galit, was incredible and so knowledgeable about all of



Synagogue in Safed

the places we visited. My photos tell part of my experience. What you can't see are the friendships made, the pride I felt for Israel, and the fulfillment I feel for having been able to enjoy this awesome gift, just as my brother Ari did last year, and other Jewish young adults have been for the past 10 years.

We boarded the plane for the return trip as a group of close friends, very tired and eager to share our experiences with our friends back home! Taking my anti-rejection meds was a little tricky because of the time difference, but I did well, and my labs after the trip were perfect!



Our group near Masada, along with leaders and soldiers who traveled with us for a few days. (I'm in front right corner)



Me at an archeological dig!



Getting more Essential Nutrients into the MSUD Diet

By Alex Larkin Bradley MS, RD

We've heard a lot about the benefits of fish and nuts lately. That's because these foods are good sources of Essential Fatty Acids (EFAs), which are known to have health benefits such as preventing heart disease and certain cancers. What's more, a deficiency of essential fatty acids can also give you dry skin in the winter! Are you getting enough in your diet?

While fats are one of the food groups that are allowed freely in the Maple Syrup Urine Disease (MSUD) diet, this very restrictive diet does not contain any meats, fish, nuts or dairy where most of us get our essential fatty acids from. So where do patients following this low protein/low leucine diet get their Essential Fatty Acids from? Are they getting enough?

Well to start with, let us try and understand what an EFA is. The word essential means that we are unable to make them within our bodies and must get them from the diet. We actually have approximately 20 different types of fatty acids within the body that have different functions, but only two of them are essential. These are linoleic acid, which is an omega-6 fatty acid, and linolenic acid also known as omega-3 fatty acid. There are presently no set Recommended Daily Allowances (RDA's) for EFA's but it is suggested that 3-6% of total daily calories come from this source. A 1:5 ratio of omega-3 to omega-6 respectively is recommended for optimal health. The typical American diet can provide a ratio from 1:10 to 1:25, and it is thought that this imbalance can cause many health problems. The MSUD diet is void of unhealthy animal fat, which is a plus, but adding more omega-3s will help even more!

Omega-6 fatty acids are readily found in a typical western diet from sources like vegetable oils such as corn, sunflower, safflower and soybean oils. Most of these oils are found in many processed foods. Good sources of Omega-3 on the other hand, are fish and nuts which are not allowed in the MSUD diet. Fortunately, flaxseed, canola, and walnut oils are also good sources of Omega-3 fatty acids and are protein free.



While other essential nutrients are provided by the MSUD formulas, EFAs are not. In fact, many of the newer formulas don't contain any fat at all. By simply adding one tablespoon of flaxseed, walnut or canola oil to your diet every day, you could prevent EFA deficiency and lead to better overall health, preventing disease and keeping your skin healthy in the winter!

Along with the beneficial omega 3 fatty acids, a diet high in fruits and vegetables is associated with a lower risk of heart disease, cancer, and obesity. Fortunately, these foods are also very low in protein and leucine, and should be the cornerstone of the MSUD diet. The basic dietary recommendation is to consume at least 5 a day of fruits and vegetables combined. Yes, that is per day not per week.

Why are fruits and vegetables so nutritious? They contain most of the vitamins, minerals, antioxidants and fiber that we need in a day including beta carotene, folate, potassium, vitamin C and vitamin A. Fruits and vegetables also contain phytonutrients, compounds found naturally in fruits and vegetables that protect against diseases such as cancers and heart disease.

Some of the most common phytonutrients that you might have heard of are lycopene found in tomatoes, watermelon and pink grapefruit, bioflavonoids found in most fruit and vegetables, and indoles found in broccoli, cauliflower and mustard greens. The different colors found in fruits and vegetables all contain different phytonutrients that all have different functions in preventing disease. Eating a variety of fruits and vegetables keeps everyone, with or without MSUD, healthier.

Start being healthier today and prevent cancers, heart disease and even dry skin by making sure you get at least 5 a day and add some flaxseed oil to your diet every day. An easy way to do this is to make sure that each meal contains either a fruit or vegetable or both. For breakfast add fruit such as blueberries to your cereal or low protein pancakes. For lunch add a side salad or a piece of fruit and at dinner have low protein pasta with broccoli, zucchini or other vegetables. Snack on an apple or a pear and carrots or celery through out the day and you already have 5 a day! It's not that hard! Add some flaxseed oil to your salad or to your pancakes to make sure you are getting your daily intake of EFA also for overall good health. ■

The Burkholder Family Story

By Verna Burkholder and edited by Joyce Brubacher

We, John and Verna Burkholder from Fleetwood, Pennsylvania, are the parents of 12 children, 6 with MSUD and 6 without. Their names are Erla (23) with MSUD, Wilmer (22), Michael (20), Lowell (18) with MSUD, Lavon (17) with MSUD, Norma (16), Duane (14), Aaron (12), Jerome (9), Luann (8) with MSUD, Joseph (5) with MSUD, Gracetta (2) with MSUD. So, as you know, dealing with the disease and diet have become part of our lives. I have read many MSUD stories and my story is similar to the others.

The disease was new for this area, for us, and for the doctors, when our first child Erla, was born. She arrived on a Wednesday, January 23, 1985 at 7 a.m. and weighed 7 lb. 8 oz. It was a normal birth, and she was a very contented baby. She even gave us a few smiles before things started to reverse.

On Saturday, I decided she was not a contented baby after all. Sunday she started taking less nourishment and was fussy. Sunday night was a night not to be forgotten. Erla had a seizure about 8 o'clock which really scared us. At the time we did not know it was a seizure. Then she seemed alright but slept poorly that night.

Erla seemed very listless in the morning, and we had the midwife come to the house—she is also our nurse. She said if Erla didn't eat all night, she should be screaming from hunger. Erla just lay on the table hardly noticing the nurse. When she clapped her hands by Erla's ear, she did not react to the sound. So she made arrangements to take her to the hospital in Reading. On the way down, Erla continued to have seizures. All day long we answered questions, and they took tests. It was very taxing to see our baby being jabbed again and again, especially since I had hardly any sleep the night before. We also started feeding her Similac with a bottle because it seemed to go better than nursing her. She took the bottle throughout the day and continued to have seizures once in a while. In the evening, they started tube feeding her. We left for home with still no answers.

I am writing this from memory, so I am not sure whether it was that evening or the next that a nurse at the hospital attended a seminar in the evening where they mentioned MSUD. This started them thinking Erla might have MSUD. The Reading hospital couldn't handle this disease, so she was transferred to Hershey Medical Center. Unknown to us, they couldn't handle it there either. So she spent a day in Hershey where they ran more tests, and, yes, she was jabbed some more. We got a glimpse of her in her incubator which still stands out in my memory. Erla had no clothes on and had wires and tubes everywhere! We left for home then as there was nothing more we could do. I don't remember exactly when or at which hospital the final diagnosis was made.

That same evening we got a call from the Children's Hospital of Philadelphia (CHOP) asking for permission to transfer Erla by ambulance because Hershey did not have the equipment to treat MSUD. So Erla was sent to the third hospital for that day. I am not sure when she went into a coma, but by the time she was in Philadelphia, she was in a poor condition. She was put on a ventilator and given medicine to control her seizures.

Erla had a long road to recovery and the doctor warned us about the possibility of brain damage. If I remember correctly, her highest leucine level was 78 mg/dl [5945 $\mu\text{mol/l}$]. She looked so sickly, both our parents came with us on the next trip to the hospital to help us make decisions as to what was best for her. By the time we got to the hospital, Erla had made the decision for us. She had made a turn for the better, and it looked more promising. I don't remember to what extent she had improved, but our hopes and spirits were lifted. They used dialysis to reduce the leucine levels. She was in the hospital a total of 3 weeks, and was released when she was 4 weeks old.

All in all, Erla is a hardy girl who rarely has the flu and has had no hospitalizations since that first one. She did have bad baby teeth that were fixed as an outpatient at the age of 2. She sat at 6 months and walked at 13 months. Her speech was delayed, and we sent her to Head Start for 2 years when she was 4 and 5 years old. She was in Special Ed. in our local Mennonite school until she finished the 8th grade at the age of 15.¹ Reading was her easiest subject, and she got to a 6th grade level in that. Her math was at a 4th grade level when she finished school. She is our helper at home and loves to wait on customers in our greenhouses and roadside

(Burkholder Family cont. on page 10)

(Burkholder Family cont. from page 9)

roadside market business. She uses the cash register and adding machine. She is also our cook and baker. Erla mixes all the formulas for herself and her 5 siblings with MSUD and sees that the younger ones drink it on time. I really miss her when she is not here for the day. In the winter months she sews comforters for Christian Aid Ministries to distribute to the poor.

Our second child is Wilmer, a healthy son born 11 months after Erla. At the age of 13, Wilmer started limping slightly and held his foot at an odd angle when biking or pushing something. To make a long story short, he had hip surgery at the age of 14 due to slipped hip joints. They put pins in, and he was out of school for a few months. He is now 22 and married.

Two years later Michael was born. He is employed by a local farmer. Twenty months later, Lowell was born and his brother, Lavon, was born 13 months after Lowell. They both have MSUD. They were tested at birth and did not need to be hospitalized. They had a good start and liked their formula. (We have not had any problems with our children accepting the MSUD formula.) With early treatment, they grew up much like normal boys. Lowell had a harder time with school lessons than Lavon. They both went to school until they were 15.1 Lowell's leucine levels in his younger years had the tendency to be on the low side and Lavon's on the higher side. We began going to the Clinic For Special Children in Strasburg when Lowell and Lavon were 10 and 11 years old, about 10 years ago. We are very thankful and feel fortunate to have the Clinic doctors, Drs. Morton and Strauss, from whom we have learned a lot over the years.

Lavon is a real farmer. Lowell, on the other hand, is an indoor person but also helps with the farm work. Lavon and Lowell both tolerate heavy farm work like any other farm boy. Lowell likes to work in our greenhouses and watch plants grow. His specialty is growing a variety of hot pepper plants. He sells the plants and peppers at our roadside market. You might also find Lowell in the kitchen cooking, especially his famous low protein donuts and chicken-like nuggets. He is left-handed and surprised his older brother Michael the other day when they decided to arm wrestle using their left hands. Lowell won.

So far, Lowell had 2 hospitalizations and Lavon 3. All of these were for the stomach virus except one of Lowell's that was due to a fungus infection in his mouth. He had to be tube fed about a week until it healed. We tube fed him at home the last 3 days.

Next in the row is Norma who does not have MSUD. How we rejoiced when we saw this baby with a head full of jet black hair. But she also had something in store for us. At the age of one week, we noticed her head was growing uneven. We had the midwife take a look at her. She was alarmed and advised us to take her to a pediatrician. X-rays showed a closed suture. Norma was diagnosed with craniosynostosis. They waited until she was 6 months, and her skull was harder and easier to work with to have surgery. The surgery was done at CHOP where our three older children with MSUD were cared for at that time. They shaved her whole head, and she had an incision from ear to ear. Her head was swollen to nearly twice its size. She was in the hospital 3 days. It didn't seem to bother her and healed up fast with no after affects.

After Norma, we had 3 boys who did not have MSUD, Duane, Aaron and Jerome. Our last 3 children have MSUD, Luann, Joseph and Gracetta. We had Dr. Morton for our doctor for these last three children. I used breast milk for their source of protein. It worked well, but I had 2 big problems—finding time and privacy! I pumped for 4 months, 2 times a day, and that supply lasted for around 8 months by freezing the extra milk. Mother's milk is lower in leucine than regular milk or Similac. I found it interesting that when the baby was in a growth spurt, it needed about half milk and half formula.

Luann (8) was a chubby 9 lb. 2 oz. baby, but she had 4 hospital stays in her first year of life. All were due to the flu. When she was 4 months old, she was very sick and had a seizure. She was hospitalized one week. Thankfully, she no longer picks up viruses easily and has had no hospitalizations since nor shown any damage from those early sicknesses. She is now 8 years old and in the 2nd grade. Luann is a little heavy for her age and always hungry.

Joseph (5) had one 24 hour hospital stay due to RSV. Gracetta (2) seems to take after Erla with no major sicknesses yet. One thing we learned was to add large amounts of isoleucine and valine to their formula as soon as they get sick. This helps tremendously to keep the leucine more stable.

Last Christmas we went to Ohio for two days. As you can imagine, it took a lot of careful planning and packing because we took all 6 children with MSUD along. My husband John's niece is Mabel Martin. She and her husband Elvin live in Ohio and have a child with MSUD also, so they supplied some of the low protein food which helped a lot. John also has 5 brothers and one sister in Ohio, and they know pretty well what the children can eat. So they had a feast of low protein foods every day. Joseph caught a virus while on the trip, so we did have to straighten out his levels when we got home.

(Burkholder Family cont. on page 11)

Heartprints
Author Unknown

Whatever our hands touch -
We leave fingerprints!
On walls, on furniture
On doorknobs, dishes,
books.
There's no escape.
As we touch we leave our
identity.

Wherever I go today
Help me leave heartprints!
Heartprints of compassion
Of understanding and love.

Heartprints of kindness
And genuine concern.
May my heart touch a
lonely neighbor
Or a runaway daughter
Or an anxious mother
Or perhaps an aged
grandfather.

Send me out today
To leave heartprints.
And if someone should say,
"I felt your touch,"
May they also sense the
love that is deep
within my heart.

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For more information or to place an order on the VitaFlo range of specialized products for Maple Syrup Urine Disease, please call 1-888-VITAFLO or visit www.VitaFloUSA.com

(Burkholder Family cont. from page 10)

Yesterday, our Luann was looking at a bottle of corn syrup. "Mom," she said, "this corn syrup has zero grams of protein and it is corn! Corn is her favorite food, and it is too bad she can have so little of it. Like I mentioned earlier, we have a roadside market business, and sometimes we wonder if we shouldn't be running a potato chip factory or a low protein noodle factory the way those disappear around here! But we are thankful there is a much greater variety of low protein foods available now than there was years ago. Our thanks to all the parents who come up with new recipes. Cooking never was my number one interest, so you can imagine that cooking two separate meals for every meal was very challenging for me.

It is time for routine blood check-ups with a finger-stick again. It is easy to push off when they are all doing well. So our normal life continues, thankfully, with many more good days than bad. We could complain that the roses have thorns, or we can rejoice that the thorn bushes have roses. Now you all take care and have a good summer. ■

1 (From page 9) Pennsylvania law states that children on a farm who attend a parochial school may quit school at 15 years of age. John's family belongs to the Old Order Mennonite church group who prefer their children do not seek education beyond the 8th grade. They teach and train their children at home. Special educational arrangements are provided for those who are through the 8th grade and are not yet 15.



Newborn Screening By Sandy Bulcher

My interest in newborn screening dates back 18 years to the birth of my second son, Jordan, on November 18, 1989. While he initially appeared healthy, by 5 days of age, he was very fussy and nursing poorly. After being treated for colic and showing no signs of improvement, he was admitted to Columbus Children's Hospital at 7 days of life. Jordan's condition gradually deteriorated from fussiness to seizure-like activity, abnormal breathing, and eventually coma. Many tests, including a spinal tap, and CAT Scan were done to determine a diagnosis without success. At 15 days of age, a geneticist informed us that Jordan was likely suffering from a metabolic disorder. After 2 more days of blood tests Jordan was eventually diagnosed with Classic Maple Syrup Urine Disease. (Incidentally, his urine did not smell like maple syrup as a newborn and has only smelled sweet several times when he was very ill.) He was 17 days of age. We were informed that Jordan probably suffered significant brain damage because of the late diagnosis. He spent several more weeks at Children's Hospital and was eventually discharged at 6 weeks of age.

Like many of you, my husband Dave and I were consumed with learning how to care for Jordan during his first year of life. Fortunately, Jordan did not experience any metabolic episodes requiring hospitalization. When Jordan was 3 years old, we changed clinics and began seeing a metabolic specialist at the University of Michigan. During his toddler years, Jordan had several hospitalizations due to metabolic crisis from illness. At a U of M metabolic conference, we met several MSUD families. I was surprised to learn that Michigan screened all newborns for MSUD and had been for several years. After further research, I learned that 20 states were screening for MSUD when Jordan was born in 1989, while Ohio was only screening for 5 disorders, including PKU, Galactosemia, Congenital Hypothyroidism, Homocystinuria, and Sickle Cell Disease. For the first time since his diagnosis, I became very angry that Jordan suffered needlessly as a result of lack of screening for MSUD in Ohio.

History of Newborn Screening

Robert Guthrie, a microbiologist and pediatrician at State University of New York, Buffalo, had a personal interest in mental retardation as his son suffered from it. Dr. Guthrie recognized that mental retardation was sometimes the result of late diagnosed PKU. In 1958, he developed a simple, inexpensive test for PKU that required only a few drops of blood. Coincidentally, Dr. Guthrie's wife's sister had a baby who was diagnosed with PKU at 15 months of age, at which point brain damage had already occurred. Dr. Guthrie became convinced that universal screening for PKU was needed and that this could be accomplished inexpensively. He began a national lecture and writing campaign about PKU and Newborn Screening. Eventually, he was given a grant to test 400,000 infants from 29 states. This study confirmed that it was possible to perform large scale testing. In 1963, a pediatric journal published Dr. Guthrie's results and parents began demanding testing for PKU. In 1963, Massachusetts became the first state to mandate PKU screening for all newborns. By 1966, a majority of states mandated testing for PKU and today all states screen for the disorder. Dr. Guthrie then turned his attention to other treatable disorders and developed tests for conditions that could be diagnosed in a newborn screening program. Sadly, Dr. Guthrie, also known as the "Father of Newborn Screening" passed away in 1995.



Ohio Newborn Screening Expansion

After learning that a screening test for MSUD in the newborn period existed and that it was mandated in other states, I began a personal campaign to see that all Ohio newborns would be screened. Initially, I met with roadblocks. A common remark that I heard was, "Mrs. Bulcher, Ohio will not screen for MSUD, because it is too rare." It didn't take me long to realize that there is strength in numbers. I assembled a group of metabolic parents and we named our organization, Ohio Coalition

(Newborn Screening cont. on page 15)



NEW FOODS by Cambrooke Foods

Toaster Topz available in two flavors is a brand new way to start your day. Just toast and spread with butter and jam. Banana Chip comes with six in a pack and have 0.1 gram Protein, 9 mg Leucine, and 160 Calories.

Tropical Topz have the wonderful flavor of pineapple and coconut, are packaged 9 to a pack and each have 0.1 gram Protein, 6 mg Leucine and 150 in Calories. These items are dry shipped. For the best flavor, place in freezer as soon as you receive them.

Welcome to chocolate paradise with the new **Fudgy Brownies**. These are a dense chocolate brownie packaged in a rectangular tray. There are 9 servings to a tray with 0.3 grams Protein, 24 mg Leucine, and 150 Calories in each.

Serve with Cool Whip and chocolate syrup or with low protein ice cream.

Asian Dumplings will add a new taste to your menu. These feature soft dough filled with onion, mushrooms, carrots, broccoli, and ginger. Pan fry till golden brown add water and simmer till all water is absorbed. There are 12 dumplings in a package with 0.3 gram Protein, 26 mg Leucine and 100 Calories per dumpling.

For a taste of the Southwest try the **New Santa Fe Biscuits**. These biscuits are filled with a southwest veggie chili. There are 4 biscuits per package, each with 1.3 gram Protein, 78 mg Leucine and 460 Calories.

These items can be ordered from Cambrooke Foods. Call toll-free, (866)-4LOWPRO, 866-456-9776 or visit their newly designed web site at www.cambrookefoods.com.

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email: orders@cambrookefoods.com
or fax: 978-443-1318.

Please send recipes to
Food News Editor

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Lemon Sandwich Cookies

- 1 cup sugar
- 1/2 cup mayonnaise
- 1 cup cold water
- 2 drops yellow food coloring
- 1 cup wheat starch
- 1 cup Cambrooke Foods Baking mix
- 2 teaspoons baking soda
- 1 teaspoon lemon flavoring
- 1/2 teaspoon salt

Preheat oven to 350 degrees. Combine all ingredients in mixer and mix well. Drop by rounded teaspoons on cookie sheet. Bake 10-12 minutes. Cool thoroughly. Makes 30 cookies. Turn 15 cookies upside down and top with 1/4 cup icing. Place second cookie on top of icing. Wrap each cookie sandwich in Saran Wrap.

Yield: 15 sandwich cookies

	Leucine	Protein	Calories
Per recipe	120 mg	2.32 g	1637
Per cookie w/frosting	8 mg	.15 g	349

Chocolate Chip Sandwich Cookies

- 1 cup sugar
- 1/2 cup mayonnaise
- 1 cup cold water
- 1 cup wheat starch
- 1 cup Cambrooke Foods baking mix
- 2 teaspoon baking soda
- 1/2 teaspoon salt
- 1 teaspoon vanilla
- 1/3 cup semi sweet chocolate chips

Preheat oven to 350 degrees. Combine all ingredients (except chocolate chips) and mix well. Fold in chocolate chips. Drop by rounded teaspoons onto a baking sheet and bake 10-12 minutes. Remove from baking sheet and cool thoroughly. Makes 30 cookies. Turn 15 cookies upside down and top with 1/4 cup icing. Place second cookie on top. Wrap each cookie sandwich in Saran Wrap. Yield: 15 sandwich cookies.

	Leucine	Protein	Calories
Per recipe	260 mg	5.32 g	1914
Per cookie w/frosting	18 mg	.4 g	367

Herb Pretzels

- 1/4 cup vegetable oil
- 1/2 teaspoon dill weed
- 1/4 teaspoon garlic powder
- 1/2 pkg. original Ranch Dressing Mix – dry
- 9 oz. broken Uncle Henry's low protein pretzels

Combine oil, dill weed, garlic powder, and dressing mix. Pour over broken pretzels. Mix well. Place in a 9"-13" baking dish. Bake at 250 degrees for 1 hour, stirring every 15 minutes. Store in an airtight container. Yield: 9 servings

	Leucine	Protein	Calories
Per recipe	171 mg	1.8 g	1390
Per serving	19 mg	.2g	367

Honey Mustard Pretzels

- 1 pound Low Protein Pretzels
- 1/2 cup vegetable oil
- 1/4 salad mustard
- 1/4 cup honey
- 2 tablespoons sour cream & onion powder

Mix oil, mustard, honey, sour cream and onion powder together. Pour over pretzels and toss to coat. Spread on baking sheet and bake at 200 degrees for 2 hours, stirring every 15 minutes. Yield: 16 1 oz servings

	Leucine	Protein	Calories
Per recipe	349 mg	4.5 g	3344
Per serving	22 mg	.28 g	209

Cambrooke Foods has recently introduced many new products - **Calzones, Veggie Meatballs, Asian Dumplings, Southwestern Biscuits, Raspberry** and **Cinnamon "Gems"**, a new line of pasta "**Solo**" and "**Duets**", as well as **Camino pro™**, our new line of food-friendly amino acid formulas for PKU and MSUD patients age five and over.

Barbecue Calzones are a fun and portable alternative to pizza. **Asian Dumplings** feature soft exterior dough filled with a savory blend of veggies and ginger with an Asian twist. The **Veggie Meatballs** are a vegetable-based savory delight rolled into meatballs. The Veggie Meatballs and Asian Dumplings are fortified with DHA Omega-3 Fatty Acids! DHA is a polyunsaturated omega-3 fatty acid that is critical for healthy eye and brain development and has also been shown to support heart health. Those on low protein diets are often deficient in DHA due to dietary limitations. Our Gems are part of a new line of cookies that are delightful bites just bursting with filling.

Cambrooke Foods invites you to 'Bring it to the Table' with **Camino pro™** our new line of food-friendly amino acid formula products for PKU and MSUD patients age five and over. The Camino pro™ line contains drinks, sauces, and sorbet stix that taste great and are designed to be enjoyed at mealtime, snack time, or on-the-go time. Bars will be coming soon.

Camino pro™ products will enhance your mealtime experience with their ability to be used in conjunction with low protein food. Mix and match **Camino pro™** easy-to-count modules to meet your protein needs. Each single-serve product is also complete with vitamins and minerals, so you never have to worry about getting all the nutrients you need! Drink flavors are Piña Colada and Fruit Punch. Sorbet Stix come in Lemon and Berry. Sauce flavors are Enchilada, Thousand Island and Balsamic Vinaigrette.

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 – as it should be. Contact **Cambrooke Foods** for a Sample Request Form.

Cambrooke Foods is ALWAYS open to serve you. 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 (2 Central Street, Framingham, MA 01701), e-mail (orders@cambrookefoods.com) or fax at (978) 443 -1318.

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MIXES THAT ARE ALWAYS AVAILABLE

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IN 5# BAGS

ALL MIXES ARE TRANS-FAT FREE.

COOKIES ARE MADE WITH NATURAL INGREDIENTS INCLUDING BUTTER
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ALL PRODUCTS ARE MADE WITHOUT ANY ARTIFICIAL PRESERVATIVES,
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Also COMING SOON.....

- COOKIE OF THE MONTH CLUB
- COOKIE BASKET OF THE MONTH CLUB
- SNACK MIXES BASKET OF THE MONTH CLUB
(MUFFIN MIX, SNACK BAR MIX, BROWNIE MIX, ETC – FLAVORS FOR MUFFIN AND SNACK BAR MIX VARY EACH MONTH)

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- ★VARIOUS PACKAGED COOKIES... APPLE-CHUNK /CRANBERRY...CHOC-CHIP ... OF COURSE.....DOUBLE CHOC CHIP.....
HAWAII MANIA.....LEMON BLAST.....CUSTOM FROSTED COOKIES.

(Newborn Screening *cont. from page 12)*

for PKU and Allied Disorders. After meeting other Ohio parents and learning about their children's disorders, my vision expanded from adding MSUD to the Ohio panel to all disorders detectable through newborn screening. Our mission to expand newborn screening in Ohio was facilitated by the addition of new technology for newborn screening, called tandem mass spectrometry (MS/MS). Prior to this, each condition required a separate test. For example, if a state tested for 8 conditions there were 8 tests. The tandem mass spectrometer allowed a state to screen for 30 disorders or more with one test. A breakthrough in our efforts to expand screening occurred in 2000 after a few parents and I met with the Ohio Director of Public Health. Our group then testified about the importance of newborn screening before the House Health Committee. Hearing the stories of Ohio children who had suffered as a result of lack of screening and meeting them personally made a huge impact on Ohio legislators, and newborn screening in Ohio began to move forward. At that time, I was also asked to serve as the parent advocate on the Ohio Newborn Screening Advisory Council. MSUD was added to the Ohio Newborn Screening panel in 2001, and today Ohio screens for 32 diseases.

Expansion of Newborn Screening Nationally

From the 1960's to the 1990's, most states screened for PKU and Congenital Hypothyroidism, along with a few other disorders. The panel of disorders varied from state to state. Decisions for adding or removing disorders in the newborn screening panel involved many complex social, ethical, and political issues. In addition, most states required that diseases meet certain criteria to be added to the panel including disorder prevalence, detectability, treatment availability, outcome, and overall cost effectiveness. In the late 90's and early 2000's, many states were experiencing rapid expansion of their newborn screening programs after obtaining MS/MS equipment. Similar to the PKU parents of the 1960's, metabolic parents became very vocal about newborn screening at this time. Several national parent organizations formed including the National Coalition for PKU and Allied Disorders and Save Babies Through Screening with a mission to improve newborn screening. Parents shared their tragic stories resulting from lack of screening, and stories of positive outcomes as a result of screening, with local and national media.

Newborn screening began to move forward on a national level. The Maternal Child and Health Bureau began

funding activities to strengthen and expand newborn screening programs. One of these projects is the Regional Genetics Collaborative. The goal of the Collaborative is for states to work together, learn from each other, and improve and equalize their newborn screening programs. In 2004, the American College of Medical Genetics (ACMG) recommended that all states screen for 29 treatable congenital conditions including MSUD. The March of Dimes has become increasingly interested in newborn screening and endorsed the ACMG recommendations of uniform screening.

Thankfully, there have been enormous improvements in newborn screening in the last 10 years. Ohio now screens for 32 diseases. Some states screen for more than 40. However, there still is not uniformity, because expansion ultimately depends on funding which varies from state to state. Despite all of the progress in newborn screening, not every state in the US tests for MSUD. According to the March of Dimes website, 44 states and the District of Columbia screen for MSUD. This equates to 95% of the 4 million babies born in the US each year being screened for MSUD. In 5 states, (Montana, Kansas, Oklahoma, Arkansas, and West Virginia) testing for MSUD is required, but the programs have not yet been implemented. Nebraska is the only state that has no law requiring MSUD screening or any MSUD screening program in place. There is still much work to be done. As new diseases are identified and new technologies are developed to diagnose more diseases, newborn screening programs will continue to expand.

Our family has been blessed in many ways. Jordan never suffered any brain damage in the newborn period, contrary to what we were originally told. He is a high school senior and has been active in the marching band and tennis team. Next fall, Jordan will be experiencing a new phase of life: college. He'll be attending one of several good schools in the Columbus, Ohio area. I've been blessed also, because I've been able to share my interest and passion for newborn screening with others. After working as a nurse in various fields of adult medicine for 25 years, I accepted a position as the newborn screening coordinator at Nationwide Children's Hospital in Columbus this past October. I am grateful to have the opportunity to educate others about the importance of newborn screening. ■

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Make your reservations now!
MSUD Symposium 2008 will be held on
June 26-28, 2008
 at the Columbus Embassy Suites in
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 See page 3 for info

This Newsletter does not attempt to provide medical advice for individuals. Consult your specialist before making any changes in treatment.